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# Communication Skills in Girls with Rett Syndrome: Perceptions of Parents, Teachers and Speech-Language Pathologists

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COMMUNICATION SKILLS IN GIRLS WITH RETT SYNDROME:  
PERCEPTIONS OF PARENTS, TEACHERS AND SPEECH-LANGUAGE  
PATHOLOGISTS

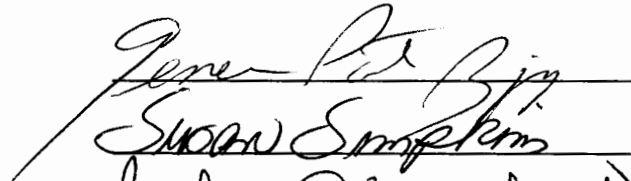
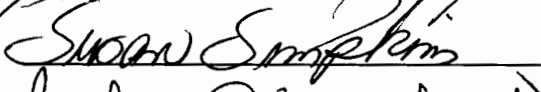

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 Date 10/13/05  
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Submitted in partial fulfillment of the  
requirements for the degree of Doctor of Philosophy in Health Sciences  
Seton Hall University  
2005

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## DEDICATION

To my family.

To my parents, Walter and Nora Edgar, for always believing in me and encouraging me to reach high.

To my husband John, for your love, support and friendship. Thank you for knowing how important this was to me, for encouraging me along the way, and for taking care of all the things that needed to get done, so that I could concentrate on this project. I could not have done it without you by my side.

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## TABLE OF CONTENTS

|   |    |
|---|----|
| ACKNOWLEDGEMENTS .....                                      | 3  |
| DEDICATION.....   | 5  |
| LIST OF TABLES.....   | 8  |
| LIST OF FIGURES.....  | 9  |
| ABSTRACT.....   | 10 |
| Chapter I.....INTRODUCTION.....                             | 12 |
| Background of the Problem.....                              | 12 |
| Pilot Study.....  | 19 |
| Purpose of this Study.....                                  | 21 |
| Research Questions.....                                     | 22 |
| Hypotheses.....   | 23 |
| Definitions.....  | 24 |
| Chapter II.....REVIEW OF RELATED LITERATURE.....            | 27 |
| Overview of Literature.....                                 | 27 |
| Description of RS.....                                      | 27 |
| Diagnostic Criteria.....                                    | 30 |
| Communication and Related Motor Skills in Stages of RS..... | 32 |
| Speech, Cognition and Apraxia.....                          | 36 |
| Summary.....  | 40 |
| Chapter III..... METHODS.....                               | 42 |
| Respondents.....  | 42 |
| Instrumentation.....  | 44 |
| Structure of the Survey.....                                | 49 |
| Data Collection.....  | 51 |
| Data Analysis.....  | 51 |
| Independent Variables.....                                  | 51 |
| Dependent Variables.....                                    | 51 |
| Statistical Analysis.....                                   | 52 |
| Analysis of Responses to Open-Ended Question.....           | 53 |

|   |     |
|---|-----|
| Chapter IV.....RESULTS.....   | 55  |
| Descriptive Statistical Analysis.....   | 55  |
| Analysis of Likert Scale Statements.....  | 67  |
| Nonparametric Statistical Analysis.....   | 74  |
| Statistical Analysis of Group Differences.....                                      | 80  |
| Analysis of Responses to Open-Ended Question Regarding<br>Communication Skills..... | 82  |
| Chapter V.....DISCUSSION.....   | 84  |
| Significant Findings.....   | 84  |
| Limitations.....  | 91  |
| Chapter VI.....SUMMARY AND CONCLUSIONS.....   | 93  |
| References.....   | 97  |
| Appendixes.....   | 101 |
| Appendix A.....Diagnostic Criteria for Rett Syndrome.....                           | 101 |
| Appendix B.....IRB Approval Letter.....   | 103 |
| Appendix C.....Solicitation Email for Participants.....                             | 104 |
| Appendix D.....Solicitation Flier for Participants.....                             | 105 |
| Appendix E..... Communication Skills in Girls with Rett Syndrome Survey.....        | 106 |
| Appendix F.....Significant Chi Square Relationships.....                            | 113 |
| Appendix G.....Themes of text responses.....  | 122 |

## List of Tables

|  |    |
|--|----|
| Table 1.....Composition of the panel of content experts.....   | 45 |
| Table 2.....Role of the Respondents.....   | 56 |
| Table 3.....Significant Differences between Teachers and SLPs.....   | 57 |
| Table 4.....Country of Residence of the Respondents.....   | 59 |
| Table 5.....Distribution of Likert Scale Scores for Perceptions of<br>Expressive Communication Skills.....   | 68 |
| Table 6.....Distribution of Likert Scale Scores for Perceptions of<br>Comprehension, Cognitive and Motor Abilities.....  | 70 |
| Table 7.....Distribution of Likert Scale Scores for Perceptions of<br>Communication Effectiveness.....   | 71 |
| Table 8.....Distribution of Likert Scale Scores for Perceptions of<br>Effectiveness of Therapy for RS, Participant Skills Level<br>in Working with RS, and Availability of Materials on<br>RS..... | 73 |
| Table 9.....Significant Differences between Perceptions of Parents<br>and Professionals in Response to Communication<br>Perception Statements.....   | 75 |
| Table 10.....Significant Associations between responses to<br>“She communicates using spoken single words” and<br>responses to other communication perception questions.....                       | 78 |
| Table 11.... Significant associations between responses to “Apraxia<br>limits her ability to communicate with others”<br>and three communication perception questions.....                         | 80 |
| Table 12.....Significant Mann Whitney U group differences.....   | 81 |



## List of Figures

|   |    |
|---|----|
| Figure 1.....Ages of girls with RS.....                               | 60 |
| Figure 2.....Communication Modalities used by Girls with RS.....      | 62 |
| Figure3.....Average Time for Girls with RS to Produce a Response..... | 64 |

## ABSTRACT

### COMMUNICATION SKILLS IN GIRLS WITH RETT SYNDROME: PERCEPTIONS OF PARENTS, TEACHERS AND SPEECH-LANGUAGE PATHOLOGISTS

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October, 2005

Rett Syndrome (RS) is a neurodevelopmental disorder that affects females and causes significant impairment in cognition, motor control and communication skills. There is little information in the literature about communication skills in RS. This study was designed to survey parents and professionals (speech-language pathologists and teachers) familiar with girls with RS about their perceptions of the communication abilities of these girls.

An online survey was developed and completed by 141 respondents (116 parents and 25 professionals) from more than 9 countries. The following hypotheses were proposed: 1) the majority of girls with RS use more than 1 communication modality; 2) eye gaze is the most frequently used communication modality; 3) respondents will agree that the cognitive abilities of girls with RS are underestimated; 4) parents will report that the girls' comprehension skills are higher relative to the perceptions of professionals; and 5) the majority of respondents will agree that motor planning difficulties impair the ability of girls with RS to communicate.

The findings of this study indicate that girls with RS are communicating, and they are perceived by familiar persons as using multiple modalities to communicate with others. Eye gaze was the modality that was consistently used by most of the girls. Apraxia was perceived as a factor in how cognition in girls with RS is assessed. Apraxia was also perceived as a barrier to successful communication in girls with RS. There was significant agreement between parents and professionals on the majority of the communication skills that were studied. The data presented here support the need for research on the use of eye gaze for communication in RS and on effective training strategies using this communication modality. Research is also needed on the nature of apraxia in this population, the impact of apraxia on cognition and communication, and how clinicians can design effective assessment and intervention strategies to address this concern.

## Chapter I

### INTRODUCTION

#### Background of the Problem

Rett Syndrome (RS) is an X-linked dominant neurodevelopmental disorder that affects approximately 1 in 10,000 females (Amir & Zoghbi, 2000). Hallmarks of the disorder are significant communication impairments, severe to profound mental retardation and poor motor skills (Gillberg, 1997). Previously considered to be an extremely rare and degenerative disorder, new information about RS has come to light in the past five years. Rett Syndrome is now known to be an X-linked genetic disorder that is caused by a mutation in the gene MeCP2 (Amir & Zoghbi, 2000; Johnston, Mullaney, & Blue, 2003). This genetic mutation was discovered in 1999, and led to improved diagnosis of the syndrome. Over 80% of cases of RS have an identified mutation in this gene (Amir & Zoghbi, 2000; Johnston et al., 2003). Long thought to be a degenerative disorder causing profound cognitive impairment, loss of ambulation skills and an early death, Rett Syndrome is now considered to be an “age-related regressive disorder of neuronal development” (Johnston et al., 2003).

The clinical presentation of girls with RS is changing as a result of diagnostic advances and greater awareness of the syndrome. Numerous studies have been published noting the wide spectrum of RS which includes girls with considerably higher cognitive abilities than previously reported (Zappella, Gillberg, & Ehlers, 1998; Zappella et al., 2003). There are reports in the literature of girls with RS who are capable of intentional communication and verbal language skills (Gillberg, 1997; Sandberg, Ehlers, Hagberg, & Gillberg, 2000; Zappella et al., 1998). These variants of RS can range from girls who display characteristics of the syndrome from birth, to girls who have far milder characteristics, and maintain oral speech. The group that has some verbal skill, known as the preserved speech variant (PSV), presents with normal head size and tends toward obesity (Neul & Zoghbi, 2004; Zappella et al., 1998). Thus girls with RS can display a wide range of abilities.

There is little published research on the communication abilities of girls with RS. The studies that do exist typically have been case reports of a single subject with RS (Evan & Meyer, 1999) or experimental designs using small groups of girls with RS (Woodyatt & Ozanne, 1993). There is a strong need for research that examines the skills of a large group of girls. Recently, a retrospective case study of 87 girls with RS syndrome was published that confirmed previous findings that girls with RS are profoundly impaired in cognitive and communication skills (Cass, Reilly, Owen, & Wisbeach, 2003).

Rett Syndrome is classified by the American Psychiatric Association as an autism spectrum disorder (ASD) (American Psychiatric Association, 1994). The term ASD was adopted to describe autism and four other similar disorders. The five individual disorders that fall under the ASD “umbrella” are Asperger syndrome, autistic disorder, childhood disintegrative disorder, atypical autism, and Rett syndrome. The disorders are related in that deficits in communication, social reciprocity and repetitive behaviors or interests are common to all, but to varying degrees. The classification of these five distinct syndromes under the ASD umbrella suggests that there is a spectrum of behaviors displayed by individuals who share key central deficits (Lord & Risi, 2000).

Girls with RS are considered to be more severely affected in the areas of cognitive and language ability than children with ASDs (Zappella et al., 1998). They are assumed to be incapable of learning to communicate intentionally or to interact socially with others (Woodyatt & Ozanne, 1992). Girls with RS are usually enrolled in educational programs because of the provisions of the Individuals with Disabilities Education Act. Since they are considered to be moderately to profoundly retarded and thus incapable of learning academic, speech or intentional communication skills, they are often not considered candidates for speech therapy or augmentative communication systems.

Yet, caregivers and teachers of girls with Rett Syndrome report that the girls can use their communication skills for a variety of purposes. There are

anecdotal reports by parents that some girls with RS use limited verbal speech, and that others use gestures or eye gaze to convey meaning. Others have been reported to learn to recognize pictures or other symbols, and some girls have been reported to enjoy stories and understand the meanings of words in books (Skotko, Koppenhaver, & Erickson, 2004).

Qualitative descriptions of skills of individuals with RS appear in several studies. In a study of 8 girls with RS, Woodyatt and Ozanne noted that all of the girls were highly social and used nonstandard behaviors to interact (Woodyatt & Ozanne, 1994). These behaviors included eye contact, vocalizations, tantrums, and body movements. This observation was confirmed in other studies (Evan & Meyer, 1999; Sigafoos et al., 2000). Persons who are familiar with girls with RS have interpreted these nonstandard movements and attributed meaning to them. However there has been concern expressed that caregivers are assigning meaning to the girls' behaviors and the girls have no expectation that the caregivers will respond (Woodyatt & Ozanne, 1992, 1993, 1994, 1997). In addition, inconsistency of response across contexts and caregivers leads researchers to questions whether the girls have clearly developed intentional communication (Sigafoos, Laurie, & Pennell, 1995).

Conversely, some research studies have indicated that when specific adaptations are made, the girls are able to display intentional behavior. In a study of augmentative communication training with three girls with RS, the

researchers demonstrated that the girls learned to touch a computer screen to make requests for desired food items (Van Acker & Grant, 1995). The respondents were between age 5 and 12 years old, and were all reported to have used meaningful speech before losing skills in the deterioration phase of RS. All 3 used communicative behaviors such as movement towards objects, gestures, and facial expressions prior to the start of the study. One subject was reported to use eye pointing (e.g. looking directly at a favored object) occasionally. During the study, the respondents were trained to touch a picture of a favored food on a computer screen. Following training, each student demonstrated increases in requesting over baseline. In addition each student demonstrated differentiated responses to "liked" versus "disliked" foods when given the opportunity (Van Acker & Grant, 1995). This study demonstrates that girls with RS are capable of learning and generalizing skills that can be used for intentional communication.

In a study that examined meaningful use of eye gaze, three girls with RS were trained to look at a named object when picture communication symbols were presented on a computer screen (Hetzroni, Rubin, & Konkol, 2002). At the end of the training, the respondents started to use the symbols during classroom instruction, which suggests that the girls learned the meanings of the symbols and were able to generalize their use to novel contexts. Though the authors cautioned that their data cannot be generalized beyond the observations made in the respondents' classrooms, they recommended that additional research be conducted to determine how to effectively train girls with RS to use symbols for



communication (Hetzroni et al., 2002). These studies provide preliminary evidence that suggests that girls with RS are capable of some level of intentional communication.

In a training study to enhance communication, Skotko et. al. (2004) noted that girls with RS could learn to communicate in meaningful ways during the context of storybook reading with their mothers. In this study, the researchers provided training to the mothers of four girls with RS. The girls ranged in age from 3.6 to 7 years old at the onset of the study. All girls had a primary diagnosis of RS and were considered to be severely communicatively impaired; they had limited to no intelligible speech, used only nonstandard gestures to communicate, and were primarily limited to vocalizations as their primary mode of communication. The mothers were trained to attribute meaning to the girls' attempts to communicate, and to ask communicatively relevant questions that the girls could answer. Mothers were also asked to wait between eight and thirty seconds to allow their daughters to respond after asking the girl a question. Reports in the literature suggest that a severe motor apraxia results in difficulty initiating movement which increases the time needed to initiate a response (Hetzroni et al., 2002; Van Acker & Grant, 1995). In the storybook reading study these adaptations resulted in increased numbers of communicative attempts by the girls with RS (Skotko et al., 2004).

The results of these studies indicate that though there is some evidence that girls with RS can communicate intentionally, there is a strong need for clinical research that describes the varied communication abilities of this population. The studies also indicate that familiar communication partners have learned to interpret the behaviors of girls with RS to determine what messages are conveyed. The role of the communication partner is key to the evolution of intentionality in the person with a severe disability (Rowland, 2003) and therefore must be further explored in order to understand the dynamic components of the relationship.

In their discussion of the evolution of intentional communication in persons with severe disabilities, Grove et. al. discuss the interdependency between the sender of the communicative act and the receiver. In an exchange where one person is severely disabled, the other partner must use inferences and intuition in order to ascribe meaning to the signal (Grove, Bunning, Porter, & Olsson, 1999). In the Skotko et. al. study (2004), the mothers were asked to assume that their daughters were competent communicators at the start of the study. As part of the training, the mothers were asked to attribute meaning to their daughters' vocalizations, eye gaze patterns and gestures. The results of the study indicated that by attributing meaning to the girls' initial behaviors, the girls were able to learn to communicate intentionally through a variety of means, including novel methods, by the end of the project (Skotko et al., 2004).

It is therefore essential to understand the perceptions of people who regularly communicate with girls with RS. When gathering data on the communication skills of girls with RS, it is crucial to obtain information about the type and effectiveness of a girl's communication ability from her communication partner. Information from a large number of partners will help build an understanding of the range of behaviors produced by girls with RS. This is a critical issue at this time as the clinical presentation of the syndrome is changing and more girls with a greater range of ability and skill level are receiving the diagnosis of RS. It is important that girls with RS are given opportunity for enhanced quality of life by developing their potential for functional communication skills.

### Pilot Study

To begin to understand the perceptions familiar communication partners have about the communication skills of girls with RS, I ran a pilot study in 2003 (Bartolotta, 2004). The purpose of the study was to interview caregivers of girls with RS to determine how they perceived these girls communicated within the context of purposeful, familiar interactions. Through open-ended questioning, I learned the modes of communication used by each girl, and what communication functions were conveyed in interactions. Two families were recruited through an electronic list serve for people with an interest in RS (the Rettnet of the International Rett Syndrome Association [IRSA]). Participants were English-speaking caregivers of medically stable daughters between the ages of 5-10

years. Each participant was interviewed for two hours in their home. The girls with RS were present and were observed interacting with their mothers during the interviews. The following themes were revealed through analysis of the transcripts:

- The respondents felt that their daughters were able to communicate a great deal of information
- The respondents believed that their daughters were unable to express much of what they wanted to communicate in a typical way because of severe apraxia
- Both families reported that their daughters were using a picture-based communication system with good reliability and success
- Both girls were reported to express their emotions, contentment, disagreement, etc. with eye gaze and facial expression
- Both families felt that their daughters' cognitive abilities were underestimated
- Both families felt frustrated by the popular belief in the field that girls with RS are significantly cognitively impaired and unable to communicate
- Both families agreed that it was difficult to read their daughters' communication signals if you were not very familiar with the girls

### Purpose of this Study

To further explore the issues raised in the pilot study, a survey was designed to collect large-scale data on the perceptions of communication skills of girls with RS by familiar persons. The target population was parents of girls with RS along with speech-language pathologists and teachers who work with girls with RS. These two groups of participants were chosen based on the following assumptions. Parents are most familiar with their daughters' skills and the unique communication behaviors displayed by each individual girl. Speech-language pathologists and teachers who work with girls with RS are experts in identifying skills and competencies in individuals with severe disabilities. These professionals are trained to assess skills in dynamic situations and are the individuals, other than the parents, most likely to be sensitive to any communication attempts made by a person with severe disabilities. They may also be more objective than parents, and because of their education and training, may require a higher standard in order to judge a behavior as communicative.

By collecting data from these two groups of respondents, I explored several issues raised in the pilot study, such as use of varied communication modality, perception of cognitive skills, and the influence of apraxia on communication. The survey was designed to collect data on the history of communication skill acquisition and current use of communication modalities in girls with RS. Respondents were asked to answer questions regarding their

perceptions of the communication skills in girls with RS. The data from the two groups of participants (parents and professionals) was analyzed to identify trends in perceptions of parents versus trained professionals. Participants were also given an opportunity to add additional descriptive information that described a particular girl's communication abilities.

In order to reach as large and representative an audience as possible, an on-line survey was designed and then publicized to parents and professionals in the Rett syndrome community around the world through email and distribution of fliers.

### Research Questions

The following research questions were posed at the start of the study:

1. What are the relationships between:
  - history of speech use and current communication skills
  - use of communication modalities and assessments of communication partners
  - method of access for pictures/symbols and comprehension abilities
  - current speech use and assessments of communication partners
2. What are the relationships between the groups of respondents:
  - how do perceptions of parents and professionals (teachers, SLPs) differ?

- how are they the same?
3. What themes do parents and professionals report when they describe the girls' communication skills?
- do respondents report that the girls' skills are underestimated?
  - do respondents report that the inconsistent behaviors produced by the girls are a barrier to educational opportunity or access to augmentative communication?
  - what kinds of strategies are parents and professionals using to enhance communication in girls with RS?

### Hypotheses

The following hypotheses were proposed:

1. The majority of girls with RS will be reported to use more than one communication modality.
2. Eye gaze will be reported to be the most frequently used modality to communicate in girls with RS.
3. Respondents will agree that the abilities of girls with RS are underestimated.
4. Parents will report that girls' comprehension skills are higher as compared to the perceptions of the professionals.
5. The majority of respondents will agree that motor planning difficulties impair the communication ability of girls with RS.

### Definitions

- 1) Rett Syndrome- a pervasive developmental disability affecting females, marked by a gradual deterioration in hand use and loss of language (Skotko et al., 2004)
- 2) Communication- "a process of encoding, transmitting and decoding signals in order to exchange information and ideas between the participants" ((Owens, 2001)
- 3) Speech-Language Pathologist- a person holding either a master's or bachelor's degree in speech pathology or a related field, whose professional work involves assessing and treating persons with communication disorders.
- 4) Intentional Communication – use of behaviors to gain another person's attention, to send a signal another person, or to seek a response from another person (Bates, Camaioni, & Volterra, 1975; Westby, 1998)
- 5) Modalities of communication – behaviors used by individuals to communicate; these may include speech, gestures, body movements, eye gaze, and writing.



6) Standard and nonstandard communication modalities used by girls with RS – these are behaviors, reported in the literature, that are used by girls with RS to communicate; these include speech, signs, gestures (pointing, head shakes), eye gaze, vocalizations, facial expressions (smiling, frowning); body movements (wiggle, kick), hyperventilation, or other (Koppenhaver, Erickson, & Skotko, 2001; Sandberg et al., 2000; Sigafoos et al., 2000; Skotko et al., 2004; Woodyatt & Ozanne, 1997).

6) Perceptions of Communication – beliefs that an individual holds regarding the purpose of a behavior.

7) Communication Perception Questions – series of statements regarding modalities of communication

8) Strong Perceptions – participant “strongly agrees” or “agrees” with a series of statements regarding communication skills of a girl with RS

9) Low Perceptions – participant “strongly disagrees” or “disagrees” with a series of statements regarding communication skills of a girl with RS

10) Communication functions – purposes of communication. In the illocutionary and early locutionary phases of communication, these purposes may involve:

efforts to regulate another person's behavior to achieve a goal; seeking of social interaction; or, to establish joint attention to share information (Bruner, 1981)

11) Apraxia - a disturbance in the ability to plan and coordinate motor movements (Hetzroni et al., 2002)

## Chapter II

### RELATED LITERATURE

#### Overview of Literature

This literature review will present current information on Rett Syndrome including the clinical features, natural history of the disorder, and the diagnostic criteria. The review will then provide an in-depth assessment of current perspectives on the communication skills of girls with RS. Three main areas will be examined: cognition and intentionality, the nature of communication attempts, and the impact of motor planning impairments on communication.

#### Description of Rett Syndrome

Rett syndrome was first described by Dr. Andreas Rett, an Austrian physician who observed two girls with similar abnormalities in his practice. Classic Rett syndrome is a clinical diagnosis that is based on defined criteria. The criteria were originally developed by Dr. Brent Hagberg and other specialists in 1985, but were revised in 2001 by an international panel of experts (International Rett Syndrome Association, 2002; Neul & Zoghbi, 2004).

## *Stages of RS*

### *Stage 1*

RS is a disorder that has four distinct stages. In the initial stage (birth – approximately 1 year), the infant typically develops normally and there is no awareness that the child has any difficulty. In Stage 1, many girls are reported to have a normal first year of life during which they begin to acquire early speech and walking skills (Cass et al., 2003).

### *Stage 2*

Sometime after the child's first birthday the symptoms of RS become evident as the girl moves into Stage 2 of the disorder. In this stage, previously acquired skills are lost, particularly motor and language skills (Naidu et al., 2003). The regression that occurs may be so slow that parents can only recognize it in hindsight (Zappella et al., 1998). Repetitive, nonfunctional hand movements begin and the child may develop seizures. In this second stage, the girl with RS typically becomes very agitated and is difficult to calm. She becomes withdrawn from social interaction and many of her behaviors are typical of autism. If the girl was verbal prior to the onset of the regressive stage, speech skills are typically lost. Development of communication and motor skills (gross and fine) usually is halted. Respiratory and sleeping disturbances become common. There is a noticeable slowing of head growth in most girls with RS during this period (Naidu et al., 2003). Scoliosis may develop in later childhood and dystonic postures of the extremities become apparent (Nomura & Segawa, 2001).

Feeding problems may also emerge in this stage. Dysphagia is common in the disorder, and problems have been reported to occur at every stage of the swallow (oral, esophageal, and pharyngeal) (Budden, 1986). These problems typically persist into adulthood. Some girls with RS are unable to feed orally and are fed through gastrostomy tubes. The risk of pneumonia secondary to aspiration is high with some children, and gastroesophageal reflux is common (Cass et al., 2003). The second stage of RS may last for 1 or more years, typically ending between ages 5 and 10 years (Cass et al., 2003).

### *Stage 3*

In Stage 3 there is typically an improvement in symptoms (Cass et al., 2003). Agitation and problems in sleeping may diminish, and the symptoms typical of Autism (i.e. loss of eye contact, social withdrawal) also seem to improve. There may be an improvement in communication and motor skills during this time. Awareness of language and comprehension usually are reported to improve and the girls may be responsive to educational programming at this time. Motor development may improve as well with gains in walking and balance typically noted (Jacobsen, Viken, & Von Tetzchner, 2001). Repetitive, non-functional hand movements, which are classic symptoms of the disorder, persist during this third stage. A girl with RS may remain in the third stage for several years. Some women with RS have been found to remain in this third stage well into adulthood (Cass et al., 2003).

#### Stage 4

Some girls with RS will move into the fourth stage of the disorder around the time of puberty. This last stage is characterized by an increasing rigidity throughout the body and a moderation of the repetitive hand movements. Eye contact is reported to improve during this stage. If a girl with RS is relatively healthy, she may live well into adulthood (Jacobsen et al., 2001). Sudden death of unknown origin, particularly during sleep, is reported in some girls with RS. There is evidence that this may be related to an abnormality in the heart rhythm. Many girls with RS have accompanying medical conditions which affect their long-term health such as pneumonia, dysphagia and seizures (Budden, 1986; Budden, Meek, & Henighan, 1990; Cass et al., 2003).

#### Diagnostic Criteria

At this time >80% of girls with a diagnosis of RS have been found to have some type of MeCP2 error (Zappella et al., 2003). The role of MeCP2 in brain development is not fully understood at this time. It is hypothesized that an error in the MeCP2 gene alters the normal developmental expression of various other genes (Amir & Zoghbi, 2000). However, diagnosis of the disorders remains *clinical*, in that a professional skilled in diagnosis of RS assesses the girl and if she is found to meet specific diagnostic criteria, the diagnosis is made. MeCP2 testing is often then performed as a confirmatory action. This involves a blood test.

The diagnostic/clinical criteria for RS were updated at a meeting of the European Pediatric Neurology Society in Baden Baden, Germany in September 2001. A panel of international experts from a variety of backgrounds convened to establish a data set to enable physicians to use uniform criteria when making the clinical diagnosis. This information was compiled based on observations and knowledge of the natural history of RS combined with new genetic information resulting from the discovery of the MeCP2 gene (Hagberg, Hanefeld, Percy, & Skjelda, 2002). There are eight essential clinical criteria which a girl must have in order to receive the diagnosis of RS. These are as follows:

1. apparently normal prenatal and perinatal history
2. psychomotor development largely normal through the first six months or may be delayed from birth
3. normal head circumference at birth
4. postnatal deceleration of head growth in the majority
5. loss of achieved purposeful hand skill between ages ½ - 2½ years
6. stereotypic hand movements such as hand wringing/squeezing, clapping/tapping, mouthing and washing/rubbing automatisms
7. emerging social withdrawal, communication dysfunction, loss of learned words, and cognitive impairment
8. impaired (dyspraxic) or failing locomotion

(International Rett Syndrome Association, 2002)

There are also eight supportive criteria that describe symptoms also displayed by girls with RS. If a girl displays some of the essential criteria, but not all, she may have atypical RS which is a milder variant of the syndrome. The entire list of supportive criteria, along with exclusion criteria and criteria for variant or atypical Rett Syndrome are contained in Appendix A.

## Communication and Related Motor Skills in the Stages of RS

### *Communication and Related Motor Skills in Stage 1*

The literature in RS contains few references to the communication skills in the first year of life in girls with RS. Anecdotal reports included in research articles note that most respondents develop early vocabularies and some develop meaningful gestures (Woodyatt & Ozanne, 1992, 1994). All reports are provided retrospectively and suggest that communication skills in the first year of life develop normally. Because RS is rarely diagnosed before two years of age, most parents are not aware that their child has a developmental disorder until well after the regression has occurred. Any retrospective data may be subject to recall bias. This is especially a problem when respondents are asked to remember information or events that are of a sensitive nature (Portney & Watkins, 2000). A clear need for data regarding the communication skills in Stage 1 of RS is evident.

### *Communication and Related Motor Skills in Stage 2*

In Stage 2 of RS there is a decline or plateauing of communication and motor skills. This regression affects hand and arm function as well as speech development. The stereotypic hand movements that the girls display take many forms. Both hands are involved, though there is typically a dominant hand that moves in a repetitive, compulsive motion. These movements are considered non-functional in that there is no clear purpose to the movement. The movements may include hand wringing, hand mouthing, or hair twirling or pulling. These



movements are characterized as compulsive because once the movements begin it seems to be difficult for the girl with RS to break the pattern and engage in other activities. Girls are reported to be able to break this stereotypic pattern and use purposeful hand movements only when highly excited or agitated. This stereotypic hand motion impacts the girl's ability to interact with and learn from her environment (Hetzroni et al., 2002; Owen, 1990).

### *Apraxia in Stage 2*

Apraxia becomes evident in Stage 2. Apraxia, which is a motor disorder of voluntary movement, can affect all planned motor movements, including limb movements and speech (Hetzroni et al., 2002). Girls with RS are believed to have apraxia of all parts of their body so that any purposeful movement is affected. Limb apraxia and apraxia of speech are especially limiting because these problems prevent girls from intentionally interacting with their environments with their hands and results in limited oral speech (Owen, 1990; Woodyatt & Ozanne, 1992, 1994). The combined effect of apraxia and the compulsive hand movements results in difficulty learning most self-help skills. In addition, this makes it especially challenging for teachers and therapists to adapt educational materials and provide access to technology.

The other outcome of apraxia is a delay in initiation of voluntary movement. It is reported that girls with RS are more likely to provide a response when their communication partner provides sufficient wait-time after asking a

question (Koppenhaver et al., 2001; Koppenhaver et al., 2001; Skotko et al., 2004).

### *Social Withdrawal in Stage 2*

The social withdrawal that is typical of autism spectrum disorders also becomes evident in the regressive phase. It is this social withdrawal in combination with limited oral speech and restricted play behaviors that results in girls with RS receiving the label of autistic disorder or atypical autism in their early years (Lord & Risi, 2000). Girls in Stage 2 of RS typically use diminished eye gaze and lose their gestures, hand waving and pointing skills (if they were previously acquired). They display limited social responsiveness in that they do not respond to their name, they do not demonstrate selective responsiveness to speech, and they fail to follow directions (Ellaway & Christodoulou, 2001; Owen, 1990).

### *Communication and Related Motor Skills in Stage 3*

Many changes take place as girls move into Stage 3 of the disorder. This occurs at approximately ages 3-5 years. The length of this stage was initially thought to be about 10 years, as it was believed that all girls then moved into stage 4, which is a destructive stage where they eventually lose their ability to walk. However new information indicates that many girls with RS stabilize at stage 3 and never progress to stage 4 (Ellaway & Christodoulou, 2001). This reason for this plateauing of symptoms is unknown at this time.

Stage 3 is a more positive and hopeful stage for families of girls with RS. During this time many girls regain some previously lost skills and also learn new skills. Some girls begin to use words in this phase. Gains are reported to occur in social interaction skills as the social withdrawal, typical of autism, begins to lessen (Zappella et al., 2003). Improvements are noted in eye contact. Many girls begin to use meaningful eye gaze to communicate messages to others. The girls are reported to be happier and more responsive to human speech. They are reported to make gains in language comprehension as well (Zappella et al., 1998).

#### *Communication and Related Motor Skills in Stage 4*

Girls with RS who progress into Stage 4 display increasing rigidity, loss of ambulation skills, decline in feeding skills, increase in seizures, and then an early demise. There is no published literature on the communication skills of women in the final stages of the disorder. However, in a multidisciplinary study of 87 respondents with RS, ages 2 years to 44 years, there was no evidence of a decline in communication or cognition with advancing age (Cass et al., 2003). As a larger number of older women are being correctly diagnosed with RS it is becoming clear that many women with RS maintain the skills from Stage 3 well into adulthood.

## Speech, Cognition and Apraxia

Speech, cognition and apraxia are three key components of the disorder. Recently researchers have suggested that these components be re-examined based on new knowledge of the disorder (Owen, 1990; Van Acker & Grant, 1995; Zappella et al., 1998). The interaction between limb and oral apraxia on cognition and language development is an issue that is not yet fully explored in this population.

Active exploration of the environment through sensorimotor activities plays a strong role in the cognitive development of young children (Owens, 2005). Children with significant physical disabilities have altered sensory experiences which affects their ability to learn from their environment (Beukelman & Mirenda, 1998). The onset of apraxia and loss of functional hand use during Stage 2 can impact a girl with RS's ability to develop cognition and language skills. Zappella et al. (1998) note that girls with RS are often initially diagnosed with only a mild cognitive impairment. It is only later, as the disorder evolves, and the motor impairments become more evident that a significant cognitive impairment is diagnosed. As RS is usually not detected in the first year of life a cognitive impairment is not visible at all during that time. Many parents report that acquisition of early milestones was unremarkable. The interaction of the evolving motor impairment on established cognitive abilities and the child's readiness to learn new skills has not been fully explored.

New information that is becoming available on the neurobiology of RS may help us clarify the cognitive changes that apparently occur as girls progress through the stages of the disorder. Enhanced excitatory activity and increased brain glucose metabolism has been found in the brains of young girls with RS who are in the early stages (1-2) of the disorder suggesting an emerging encephalopathy. Reduced excitatory activity and relatively lower brain glucose metabolism have been found in older girls who, in the later stages 3-4, are demonstrating severe cognitive impairment (Johnston et al., 2003). As more information is collected on the effect of MeCP2 mutation on the brains of girls with RS we should gain a greater understanding of the resulting influence on behavior.

The influence of apraxia on the ability to measure current levels of cognition and communication skills has been discussed by several researchers (Evan & Meyer, 1999; Sandberg et al., 2000; Woodyatt & Ozanne, 1992, 1994). Evan and Meyer studied the communication skills of an 8-year-old subject with severely impaired motor skills and RS. Their subject had failed to learn sign language or oral speech. The researchers observed this girl in a variety of interactions and noted that she used many nonstandard behaviors that were communicative. For example, she moved towards an object and looked at it when making a request. The researchers concluded that this child was capable of intentional communication using nonstandard communicative behaviors (Evan & Meyer, 1999).

Woodyatt and Ozanne (1994) concluded that apraxia is likely to interfere with a child's attempts to communicate and also results in an underestimation of the child's linguistic and cognitive abilities. These authors studied four girls with RS using a multiple case study design. They concluded that three of their respondents were at a preintentional level of communication. However all of their respondents were highly social and responsive to speech. They all used nonstandard communicative behaviors to interact with others. Examples of these behaviors are touch, eye gaze, smiling, vocalizing, and laughing. The authors suggested that future studies be designed to investigate the interrelationships between cognition, communication and physical skills (Woodyatt & Ozanne, 1994).

In a study of the use of AAC training with three girls with RS, Van Acker & Grant (1995) found they were able to train their respondents over a 3-month period to touch a computer screen to make requests for desired food items. As they studied their respondents, these researchers noted that the girls displayed a delayed motor response and required intense motivation to initiate purposeful movement. These authors suggested that the motor deficits displayed by the girls with RS may have been more severe than the cognitive deficits. The subject in the study who had the lowest cognitive level, as measured by the Uzgiris and Hunt Ordinal Scales of Infant Development (Uzgiris & Hunt, 1975), learned the skill of touching the computer screen to make a request more rapidly

than the other two respondents. All three of the respondents were reported to have cognitive abilities below Stage V of the Piagetian stages of sensorimotor development which would suggest that they were preintentional. However, the authors reported that their respondents did display intentional communication as evidenced by their ability to use the computer in a meaningful and consistent manner. In addition, they noted that the respondents displayed unique behaviors (i.e. eye gaze, vocalizations, facial expressions, gestures, walking to a desired object or activity) that were communicative in nature (Van Acker & Grant, 1995). This observation is noted in other studies as well, confirming the suggestion that these behaviors are meaningful and should be valued as intentional communication attempts (Evan & Meyer, 1999; Sigafos et al., 2000; Skotko et al., 2004)

Another skill that has not yet been explored is the failure of girls with RS to develop imitation skills. Imitative behavior is a key skill that is important in early learning especially in the first two years of life (Owens, 2005). It is unclear whether the failure to imitate the speech or motor acts of others is due to an apraxia or a lack of motivation, which would arise from a cognitive-linguistic impairment (Woodyatt & Ozanne, 1992).

Girls with RS have been noted to produce delayed responses when presented with stimuli (Koppenhaver et al., 2001). This delay in response time may arise from apraxia. A child with whole body apraxia would respond in a

much delayed fashion to a command or other type of sensory input. Researchers have noted that the girls with RS in their studies showed a temporal delay in responsiveness (Evan & Meyer, 1999; Van Acker & Grant, 1995). In an intervention study that involved parent training, it was demonstrated that providing sufficient wait-time and support after asking a question resulted in increased communicative attempts by girls with RS (Skotko et al., 2004).

### Summary

It is evident that there is growing evidence that some girls with RS display intentional communication that is recognized by familiar communication partners. There is a wide disparity in reports of cognitive potential of these girls. The influence of a motor planning disorder such as apraxia has not yet been determined and the evidence at this time is only speculative. There is a clear need for a large-scale study to collect evidence on the types of communication behaviors girls with RS currently display. As discussed earlier in this literature review, it is important that familiar communication partners such as caregivers or parents be used as reporters as they are best able to judge each individual girl. However, in order to limit potential overestimation of communication ability it will be important to also gather data from professionals who are trained to objectively evaluate nonstandard communication behaviors in informal contexts.

Therefore a survey was designed to be completed by a parent or a caregiver of a girl with RS and also by a speech-language pathologist and



teacher who is currently working with a girl with RS. The survey was used to collect quantitative data on the communication skills of girls with RS from the perspective of both the parents/caregivers and professionals (speech-language pathologists and teachers) who work with girls with RS and are familiar with their skills and communication attempts.

## Chapter III

### METHODS

This study used a between respondents, descriptive survey design to study the perceptions of the communication skills of girls with Rett Syndrome (RS) by parents, speech-language pathologists and teachers. In order to achieve a medium effect size of .30, a minimum of 87 respondents were required in order to obtain a power of .80 with one degree of freedom at the .05 level (Portney & Watkins, 2000).

The Seton Hall University Institutional Review Board (IRB) approved the research proposal on March 30, 2005. A copy of the letter from the IRB is contained in Appendix B. A total of 141 participants were included in the study; 116 were parents of girls with RS, 11 were teachers, and 14 were speech-language pathologists working with a girl with RS.

#### Respondents

Participants were recruited for the study using 2 methods. One method was by email posted on the Rettnet, an electronic listserve administered by the

International Rett Syndrome Association (IRSA). Permission to post this solicitation email was granted by Kathy Hunter, Founder of the IRSA. This listserve has approximately 600 members, consisting of parents and family members of girls with RS and professionals who work in educational or health arenas with this population. The members of this listserve reside in multiple countries from many diverse regions in the world. A solicitation email was sent to members of the Rettnet, describing the purpose of the study, and inviting interested participants to log on to the URL where the survey was stored. Specific inclusion criteria were: 1) participant must be a parent or caregiver of a girl with RS; or 2) participant must be a teacher or speech-language pathologist (SLP) currently working with one girl with RS. A sample of this email is contained in Appendix C.

The other method of solicitation involved distribution of fliers describing the study. A sample of the flier is included in Appendix D. This flier described the purpose of the study, identified the participants who could complete the survey (parents, teachers or SLPs), and contained the URL where the survey was located. The fliers were distributed by hand to attendees of the Annual Meeting of the International Rett Syndrome Association in San Antonio, TX from May 27-30, 2005. Permission to distribute the fliers was obtained from the administrative office of IRSA. Fliers were also hand-delivered and mailed to schools for children with disabilities in New Jersey and Pennsylvania that were known to be attended by girls with RS.

The sample population consisted of parents, teachers and SLPs in the target population who voluntarily agreed to participate in the study after receiving the email notification or the flier. The email and flier were written directly to potential participants and explained the purpose of the study. In addition, the protections used to safeguard their confidentiality and identities were described. Consent was demonstrated by the participant voluntarily completing the on-line survey (Portney & Watkins, 2000).

#### Instrumentation

Validity studies were conducted on a preliminary survey. A panel of seven experts assessed the content and face validity of the preliminary survey. These individuals all demonstrated significant expertise in the field of Rett syndrome and communication. See Table I for the professional background of the panel. Two members of the panel were from Australia, and the remaining 5 were from the United States (Maryland, North Carolina, New Jersey, Ohio, and Illinois).

Table I

## Composition of the Panel of Content Experts

| Number | Professional Background         |
|--------|---------------------------------|
| 1      | Parent, Founder of the IRSA     |
| 2      | Professors of Special Education |
| 3      | Speech-language pathologists    |
| 1      | Epidemiologist                  |

This panel of experts was asked to review the preliminary survey and provide recommendations on the wording of items, deletion or addition of any items, and the overall style and organization of the survey. An item-by-item analysis was conducted to assess percentage of agreement among the panel of experts. The researcher initially reviewed all recommended content, design and organizational revisions. Appropriate content revisions involving wording of items, and alteration of any items (including additions) were incorporated. Based on feedback from three experts, the survey (which was initially to be sent to parents only) was modified to also be sent to teachers and SLPs. The recommendation was that teachers and SLPs who are familiar with RS are expert in interpreting the communicative behaviors in girls with RS, and may provide alternative perceptions relative to the parent group. In addition, the items pertaining to judgment of communication behavior were reworded. A series of

statements regarding communication skills were designed so that participants could rank their response along a five-point Likert scale. A Likert scale is a summative scale that is used to measure attitudes or opinions (Portney & Watkins, 2000). Use of the Likert scale enabled statistical comparisons to be made between the groups of participants.

The revised survey was then sent to the panel of 7 experts for a final review. No other substantive recommendations were offered during this second review phase. A pilot test phase was then introduced to ensure that professionals would have sufficient background information on girls with RS in order to completely fill out the survey. The survey was distributed to the Director of Speech-Language Pathology services at a school for children with multiple disabilities in New Jersey. This school had at least ten students with RS enrolled at the time of the study. Three teachers and three SLPs, all working with girls with RS, volunteered to complete the survey anonymously. Results were analyzed as follows: 1) willingness of potential respondents to participate in the study; 2) comprehensibility of questions; 3) relevance of questions; and 4) variability of responses to the communication perception questions.

All participants completed the survey independently and did not write any comments regarding difficulty understanding or completing any survey item. Four of the participants completed every item. One participant did not complete a question in the demographic section asking whether the girl with RS ever spoke

or used words. One participant did not answer one of the communication perception questions.

Analysis of answers to the demographic questions, which obtained background information on the girls with RS, indicated that 3 of the 6 participants (50%) replied “don’t know” to the question that asked what stage of RS the girl was in currently. According to a recent publication, the stage of RS has not been found to significantly correlate with level of language acquisition (Cass et al., 2003). Since it is likely that most teachers and SLPs will not know which stage a girl with RS is currently in, and there is little information that suggests it is relevant for perception of communication skills, that question was deleted from the final version of the survey. All 6 participants answered the question about whether the girl with RS had an identified MeCP2 deletion as “don’t know”. Since it is likely that only the parent participants will know if a MeCP2 deletion has been identified in a girl with RS, that question was also deleted from the final version of the survey.

The responses to the perception of communication skills questions were distributed across all 5 choices. Out of a total of 119 possible responses, the percentages of responses within each Likert category were as follows: 18% “strongly agree”; 36% “agree”; 13% “undecided or unsure”; 21% “disagree”; 12% “strongly disagree”. This pattern of responses was expected and affirmed the

types of questions asked in the survey, and helped shape the hypotheses that were posed before data collection began.

The third section of the survey provided the participants with an opportunity to share any additional information by responding to an open-ended question regarding the girl with RS and her ability to communicate. Four of the 6 participants (2 teachers, 2 SLPs) in the pilot study supplied responses in this section. They are as follows:

"I feel that she understands more than people think she does, and more than she is able to communicate to us".

"The girl with Rett syndrome that I work with has made great gains in regard to picture identification in a field of two. She has also communicated her feelings of affection for the teenaged boys in the class by reaching, smiling and vocalizing".

"My little girl chooses via pointing with her chin. She primarily makes choices. She does not discriminate between multiple pictures".

"I believe that eye gaze is an important mode of communication for girls with RS and is often under used. I also have found their ability to communicate under-estimated by therapists and the educational community. The girls seem to



thrive when they a (are) familiar with the individuals working with them and frequent change (is) detrimental to their progress”.

The themes expressed in these comments provided support for keeping this section in the final version of the survey. Given the complex and dynamic relationships between girls with RS and their communication partners, it was suspected that the responses to the open-ended question would provide a rich source of information to support the quantitative data.

### Structure of the Survey

The final version of the survey entitled “Communication Skills in Girls with Rett Syndrome” is contained within the Appendix E. The survey consisted of three sections: (1) demographic information; (2) perceptions of communication skills; and (3) additional descriptive information. In the demographic information section, participants were asked to identify themselves as either a parent of a girl with RS or a teacher or SLP working with a girl with RS and where they resided (in a section of the United States, one of 14 other countries, or in an unnamed country). A total of 13 questions specifically relating to the girl with RS were included in this section. The following information was collected: the age of the girl, what communication modalities she used, the access method utilized if she used pictures/symbols to communicate, the average time required for the girl to produce a response, history of speech and related therapies, current educational

placement, history of acquisition or loss of verbal speech skills, and whether the girl used any verbal speech at the time of the survey were included.

The second section contained a series of 20 statements regarding the communication skills of the girl with RS. There were four groups of statements: expressive communication skills (8 statements), comprehension, cognitive and motor abilities (5 statements), communication effectiveness (4 statements), and 3 statements regarding the availability of materials on RS, participant knowledge of RS and the effectiveness of therapy. A five-point Likert scale was used. Participants were instructed to read each statement and choose one of the following five choices based on their perception of the girl with RS's communication skills: strongly agree, agree, undecided or unsure, disagree, strongly disagree.

The third section provided the participants with an opportunity to share any additional information about the girl with RS and her ability to communicate. Given the dynamic nature of communication, this section was considered to be crucial to the survey. This section provided qualitative information to support the quantitative data collected in section two. The results of the pilot study revealed that open-ended questions provide a great deal of individual information regarding each participant.

### Data Collection

The survey was housed on the Seton Hall University's secure server through the ASSET: Academic Survey System and Evaluation Tool project available through the Department of Information Technology. The participants were required to log on to the survey at a unique web address, and then enter a user name consisting of the first three letters of their first and last names. This was done to ensure that each participant accessed the survey once, reported their perceptions about an individual girl, and that confidentiality was maintained. The survey was available for access from May 1, 2005 to June 30, 2005.

### Data Analysis

Data obtained from each of the 141 submitted surveys were coded and downloaded to the Statistical Package for Social Sciences (SPSS 12.0) for data analysis. The data was examined in order to determine which statistical methods would be most appropriate for describing the demographic information, and analyzing the relationships between the independent variables and the dependent variables.

### Independent Variables

The fifteen independent variables were the answers to the demographic questions, described as follows: (1) role of participant [parent, teacher or SLP]; (2) country of residence; (3) age of girl; (4) communication modalities used; (5) access methods; (6) time to respond; (7) therapies received; (8) educational

enrollment; (9) type of educational setting; (10) history of speech use; (11) history of speech loss or regression; (12) age of loss/regression of speech; (13) amount of speech used prior to regression; (14) use of speech currently; (15) specific words or phrases used currently.

### Dependent Variables

The dependent variables were the responses to the 20 Likert scale statements. Each statement had five levels of responses: (1) strongly disagree; (2) disagree; (3) unsure or undecided; (4) agree; (5) strongly agree.

### Statistical Analysis

Data analysis included both descriptive and nonparametric statistics. Descriptive statistics allow a researcher to describe the central tendency, and variability with a set of data in order to describe a population (Portney & Watkins, 2000). The data collected in the survey included both nominal and ordinal data. For nominal data, percentages, standard deviation, mean, and mode are valuable statistics to describe central tendency. For ordinal data, which is the data collected in response to the Likert scale statements, the median is a valuable measure of central tendency to report in addition to the mode (Nardi, 2003). Therefore, for the descriptive analysis, frequencies, median, mode, mean, standard deviation and percentages were reported.

For the nonparametric statistical analysis, the Chi Square Test of Homogeneity ( $\chi^2$ ) was selected to study relationships between the independent and dependent variables using cross-tabulations (Nardi, 2003). Cross-tabulations are a common approach to data analysis for describing relationships between two or more sets of responses (Portney & Watkins, 2000). To study the strength of the relationships between the variables, Kendall's Tau-b ( $\tau$ ) was selected (Nardi, 2003). Kendall's Tau-b is a measure of association for ordinal or ranked variables where data is reported in 2 x 2 tables. The Kendall's statistic is interpreted in terms of direction (positive or negative relationship) and strength (weak to strong). The sign of the coefficient indicates the direction of the relationship, and its absolute value indicates the strength, with larger absolute values indicating stronger relationships. Possible values range from -1 to 1 (Nardi, 2003; Portney & Watkins, 2000). The Mann-Whitney U Test was selected to study the differences between the two groups (parents and professionals) of respondents. This test is appropriate when analyzing ordinal data, and is used to compare data from two groups (Portney & Watkins, 2000). An alpha level of  $p \leq .05$  was considered significant for all statistical tests (Nardi, 2003; Portney & Watkins, 2000). This is an appropriate level to set for this type of exploratory research (Nardi, 2003).

#### Analysis of Responses to Open-Ended Question

The respondents' responses to the open-ended question in section three of the survey were analyzed using qualitative research techniques (Strauss &

Corbin, 1998). Themes expressed by the respondents, using keyword analysis, were identified. The statements expressed by the respondents were analyzed and compared to the quantitative data. Impressions developed from these statements will be used to frame future research questions and contexts.

## Chapter IV

### RESULTS

A total of 152 persons attempted to complete the survey. Eleven (11) of the surveys were incomplete at the time that the survey was closed on June 30, 2005, so those responses were discarded. This resulted in a total of 141 completed surveys available for analysis.

#### Descriptive Statistical Analysis

Demographic characteristics of the persons completing the survey (respondents), their country of residence, and the age of the girl with RS they reported on in the survey, are displayed in Tables 2-4 and Figure 1. The role of the respondent (parent, teacher or SLP) and the number in each group is presented in Table 2.

Table 2

## Role of Respondents

| <b>Role</b>         | <b>Number</b> | <b>Percent</b> |
|---------------------|---------------|----------------|
| Parent or caregiver | 116           | 82.3           |
| Teacher             | 11            | 7.8            |
| SLP                 | 14            | 9.9            |
| <b>Total</b>        | <b>141</b>    | <b>100.0</b>   |

*Role of Respondents*

Since the individual groups of professionals (teachers and SLPs) were fairly small relative to the size of the parent group, the responses from the teachers and SLPs were analyzed to determine if any significant differences existed between the two groups. A Mann Whitney U test was conducted on the responses to the 20 Likert scale statements. Significant differences were found in responses to 2 of the 20 statements. The data is presented below in Table 3.



Table 3

Significant Differences between Teachers and SLPs

| Statements  | Mean Ranks |       | Asymp. Sig. (2-tailed) |
|---|------------|-------|------------------------|
|   | Teachers   | SLPs  |                        |
| She uses body movement to communicate                               | 9.55       | 15.71 | .026                   |
| Her communication skills have improved with training and/or therapy | 9.41       | 15.82 | .020                   |

These findings indicate that although teachers' & SLPs' perceptions of girls were overwhelmingly similar, the groups did vary significantly in their perceptions for 2 aspects. SLPs were more likely than the teachers to agree that girls with RS used body movement (e.g. kicking, moving to an object) to communicate. In addition, SLPs were more likely than the teachers to agree that the communication skills of a girl with RS had improved with training and/or therapy. These differences, though notable, were considered to be small enough to allow the data from the 2 groups of professionals to be combined to represent 1 group.

Though teachers and SLPs certainly have different levels of education with regard to interpretation of communicative behavior, these professionals who work with the multiply disabled population must learn to interpret the individual behaviors of the students (clients) they work with and teach on a daily basis. Therefore it made clinical and statistical sense to combine to data from the teachers and SLPs to represent the professional group. This group of professionals as a whole would likely be fairly different from parents, most of whom do not have formal training in education of people with significant disabilities, such as RS.

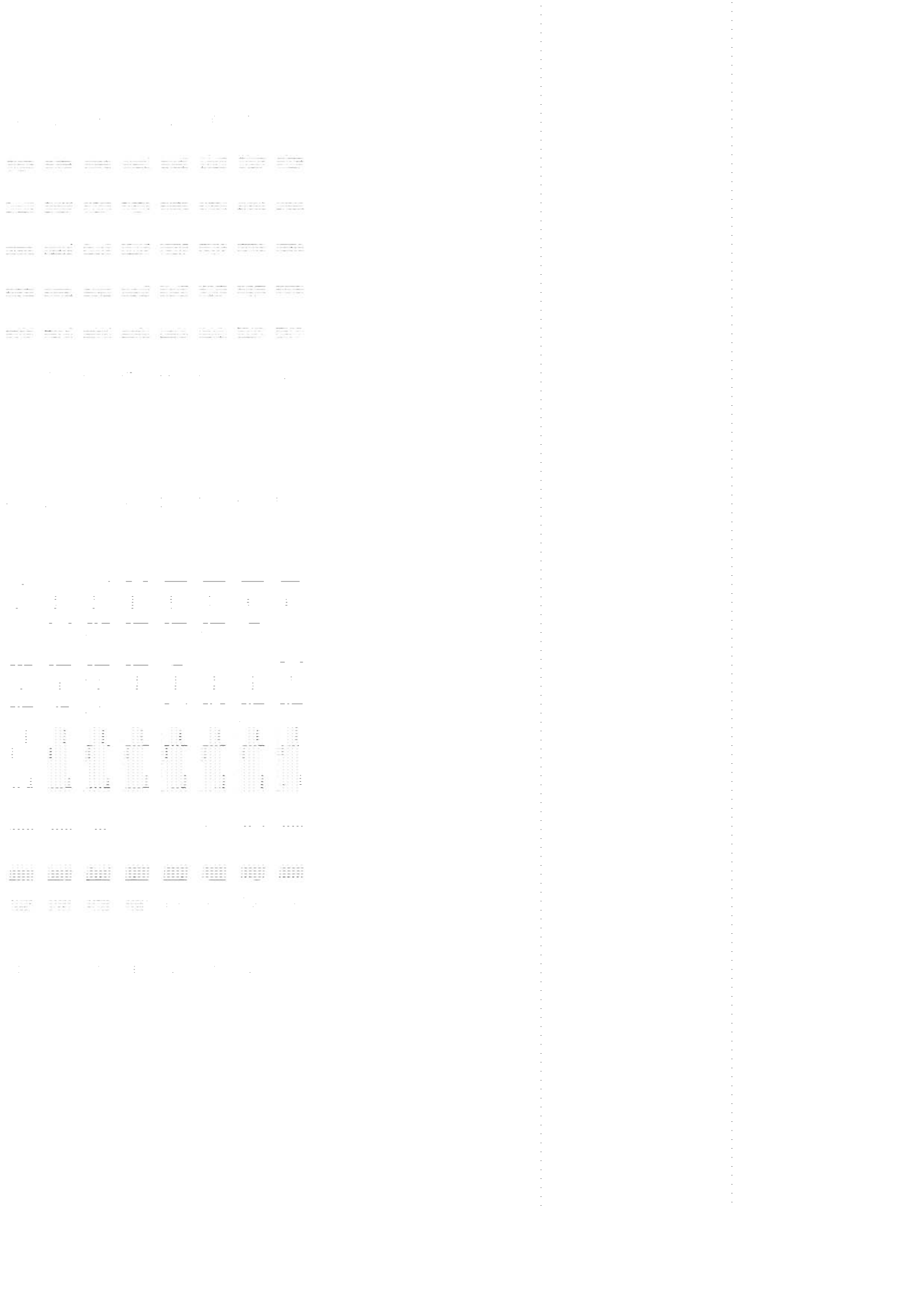
#### *Country of Residence*

The data on country of residence is presented in Table 4. The majority of the respondents (83.7%) resided in the U.S. The other respondents were from at least seven other countries, including Australia, England, Ireland, Israel, Italy, Portugal, and Scotland. A total of eleven respondents reported that they lived in countries other than the fifteen listed in the survey. At least one of these respondents resided in Canada, based on information they provided in section three of the survey. In summary, 118 of the respondents lived in the U.S., with the remaining 23 respondents (16.3%) distributed across at least eight countries.

Table 4

## Country of Residence of the Respondents

| <b>Country (or area of U.S.)</b> | <b>Number</b> | <b>Percent</b> |
|----------------------------------|---------------|----------------|
| Northeastern United States       | 72            | 51.1           |
| Southeastern United States       | 16            | 11.3           |
| Northwestern United States       | 17            | 12.1           |
| Southwestern United States       | 13            | 9.2            |
| Australia                        | 4             | 2.8            |
| England                          | 3             | 2.1            |
| Ireland                          | 1             | .7             |
| Israel                           | 1             | .7             |
| Italy                            | 1             | .7             |
| Portugal                         | 1             | .7             |
| Scotland                         | 1             | .7             |
| Other                            | 11            | 7.8            |
| <b>Total</b>                     | <b>141</b>    | <b>100.0</b>   |



Handwritten musical notation on a single staff, featuring a treble clef and a key signature of one flat. The notation includes a series of eighth and sixteenth notes, with some measures containing rests. The handwriting is in a cursive style, typical of 18th or 19th-century manuscripts.

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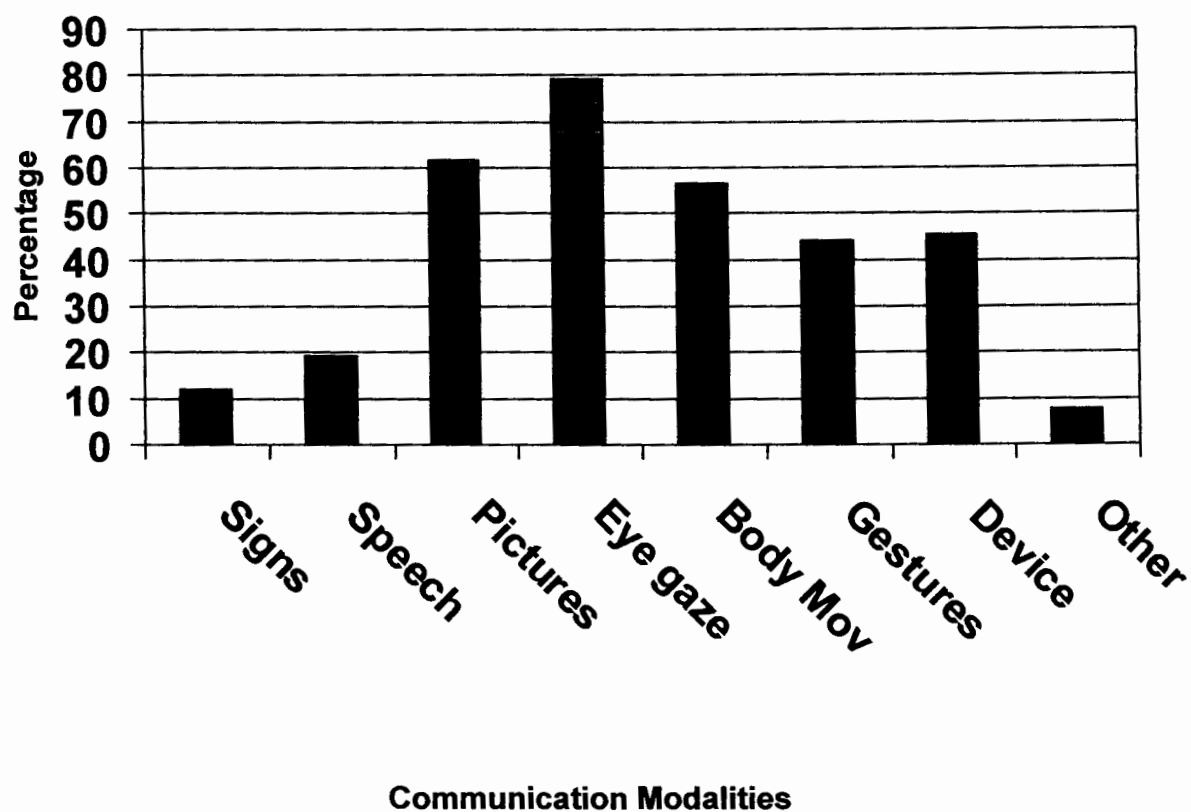


### *Communication Modality Use*

All respondents answered this question. They reported that most girls with RS used more than one modality to communicate. There were 8 levels of responses for this survey question. The choices were: signs, verbal speech, pictures/symbols/objects, eye contact/gaze/pointing, body movement, gestures, voice output device, or other unspecified modality. The data indicated that each girl uses an average of three different modalities to communicate information to others. Figure 2 presents the distribution of the data across various communication modalities. Eye gaze in some form was reported to be the most frequently used communication modality, as it is used by 78.9% of the girls with RS. Body movement and pictures or symbol boards were the next most frequently reported communication modality, used by 56.3% and 61.3% of the girls with RS, respectively.

Figure 2

Communication Modalities Used by Girls with RS





### *Access Method for Communication Systems*

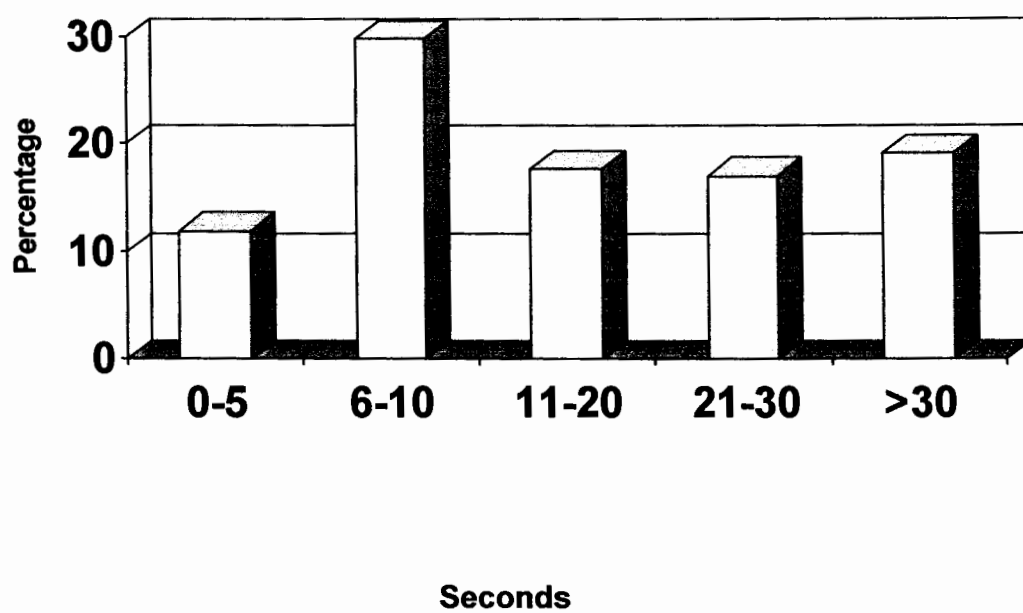
The next question in the survey concerned the method that girls used to access their communication systems. There were five levels of responses from which the respondents could choose. Interestingly, for those girls who are able to access a communication system, 60.3% use touch with hands or fingers and 57.4% use some type of eye gaze. A head pointer was reported to be used by only 2.8% of girls, and other types of access (not specified) were used by 5% of the girls. Only 11.3% of the girls with RS were reported not to use a communication system.

### *Response Time*

A total of 134 of the 141 respondents responded to the question "On average, how long does it take for her to generate a response?" (Figure 3). The length of time required for girls to generate a response varied across all five response levels. The largest number of girls with RS (n=42), which represents 31.3% of the responses, are reported to need between 6-10 seconds to generate a response. Approximately 12% of the girls generate a response in 5 seconds or less, while 19 % respond in 11-20 seconds and 18% generate responses in 21-30 seconds. A total of 27 respondents reported that a girl needed more than 30 seconds to generate a response. This number represented approximately 20% of the total population. In summary, this data indicates that 57% of the entire group of girls required a delay of 11 seconds or more to generate a response.

Figure III

Average Time for Girls with RS to Produce a Response



### *Therapy History*

Over 90% of the girls with RS have received speech, physical and occupational therapies. Fifty percent have received an augmentative communication evaluation. Just over one-third (38%) have received music therapy. A total of 3 girls (2.1%) have not received any of these therapeutic services. Two of the girls reside in the U.S. and 1 is in Portugal. The girl in Portugal is between 4-7 years of age. One U.S. resident is from the Northeast and is between 13-20 years of age and the other is from the Northwest and 21 years of age or older.

### *Educational Programming*

Over 84% of the total girls are currently enrolled in some type of educational program. At least 61% are in a special education classroom. A total of 24 girls (18%), are included in a regular class with typical peers. Six of the girls (4.5%) are in an adult day program. Eleven of the girls are not in any type of program at all. Two of the respondents did not answer this question.

### *History of Speech Use*

Seventy percent of the girls are reported to have used oral speech at some time in their life. Twenty-five percent have never spoken, and seven respondents were not sure about a specific girl's history of speech use. Two respondents did not answer this question.

Of the girls who were reported to speak orally at the time of the survey or at some time in the past, 86% have experienced a loss or regression in speech skills. For 59%, this regression occurred between 1 and 2 years of age. Approximately 11% were less than 1 year of age at the time of regression, 20% were between ages 2 and 3 years, and 10% were older than 3 years of age. Forty-four percent of the girls who lost speech during a regression had less than 5 single words before the regression occurred. Thirty-seven percent had acquired between 10 and 50 single words, 11% used short phrases, and 6% had developed sentence use before the regression. Of the six girls who were reported to have acquired sentence use before the regression, three of them were older than 3 years when the loss of speech occurred. The remaining three girls were between 1 and 2 years ( $n=1$ ) and between 2 and 3 years ( $n=2$ ) of age at the time of speech loss. Of those who were reported to have a history of speech use, 46% are reported to speak at the current time.

Seventy text responses were submitted in response to the question "What words or phrases does she say". The data indicates that 69 of the girls are using some type of verbal speech. The range of abilities reported was quite large. Eleven girls are reported to use one word only at this time. Two girls are reported to have near normal speech; one girl speaks in 5-6 word utterances; one girl has over 100 hundred single words and can use them in phrases beginning with "I want.." or "need a ..."; and another girl is currently using approximately 100-125 words and some short phrases. Most girls ( $n=56$ ) are

reported to use between one and ten words. The most commonly reported word (n=37) was a version of mother (ma, mom, mommy).

### Analysis of Likert Scale Statements

Section two of the survey contained 20 Likert Scale statements that measured the respondents' perceptions of the girls' communication skills. Descriptive statistics, including mean, standard deviation, median and mode are reported for each question and presented in groups (described earlier in Method section) in Tables 5-8. This analysis allowed the researcher to determine the level of agreement or disagreement for each statement and to construct impressions about the respondents' overall perceptions of the communication skills of girls with RS. All 141 respondents provided a response for each statement.

Table 5 presents the distribution of the Likert scale scores for the perceptions of the expressive communication skills statements. There were three questions in this section for which the mode was 5 (strongly agree). These questions were related to communication using eye gaze, vocalizations, and body movement. Fifty-seven percent of respondents chose "strongly agree" as a response to the eye gaze question. Forty-six percent chose "strongly agree" for

the vocalization question, and strongly agree was chosen by 37% of respondents for body movement.

Table 5

Distribution of Likert Scale Scores for Perceptions of Expressive Communication Skills

| Statement              | Mean | Standard Deviation | Median | Mode |
|------------------------|------|--------------------|--------|------|
| Communicates using.... |      |                    |        |      |
| spoken single words    | 2.09 | 1.43               | 1      | 1    |
| spoken sentences       | 1.41 | 0.90               | 1      | 1    |
| vocalizations          | 4.02 | 1.30               | 4      | 5    |
| sign(s)                | 1.87 | 1.15               | 1      | 1    |
| pictures or symbols    | 3.65 | 1.40               | 4      | 4    |
| gestures               | 3.12 | 1.50               | 4      | 4    |
| eye gaze               | 4.33 | 0.99               | 5      | 5    |
| body movement          | 3.79 | 1.34               | 4      | 5    |

In summary the data presented in Table 5 indicate that for the expressive communication skills statements, the respondents perceived that most girls with RS communicate with eye gaze and vocalizations and body movements, and far fewer girls use spoken sentences and signs.

The distribution of the Likert scale scores for the perceptions of comprehension, cognition, and motor abilities statements is presented in Table 6. There were two questions in this section for which the mode was 5 (strongly agree). The respondents demonstrated the strongest agreement with the statement that girls with RS understand at least 10 spoken words. Seventy-three percent of respondents chose strongly agree for this question. Strong agreement with the statement that apraxia limits the ability to communicate was also evident based on the data. The response choice of strongly agree was chosen by 58% of respondents. Of note is that the standard deviation for each of these questions was fairly small ( $<1$ ) indicating little variability in responses of respondents to this question.

Table 6

Distribution of Likert Scale Scores for Perceptions of Comprehension, Cognitive and Motor Abilities

| Statement                                 | Mean | Standard Deviation | Median | Mode |
|---|------|--------------------|--------|------|
| Can read 1 or more single words           | 2.66 | 1.52               | 3      | 1    |
| Understands at least 10 spoken words      | 4.60 | 0.76               | 5      | 5    |
| Follows 1-step commands                   | 3.79 | 1.18               | 4      | 4    |
| Has a significant cognitive impairment    | 2.93 | 1.28               | 3      | 4    |
| Apraxia limits her ability to communicate | 4.35 | 0.91               | 5      | 5    |

In summary, the data presented in Table 6 indicate that for the other comprehension, cognition and motor abilities statements, the respondents perceived that most girls with RS understand at least 10 words and that apraxia is a significant barrier to communication.

The distribution of the Likert scale scores for the perceptions of communication effectiveness statements is presented in Table 7. There were two questions in this group for which the mode was 5 (strongly agree). Most respondents agreed with the statement that familiar people can interpret the communication of a girl with RS; the mean was 4.41. Fifty-five percent of



respondents chose strongly agree for this statement. Respondents were also likely to strongly agree with the statement that most people underestimate the abilities of girls with RS; the mean for this statement was 4.30. Sixty-five percent of respondents chose strongly agree as their response choice.

Table 7

Distribution of Likert Scale Scores for Perceptions of Communication Effectiveness

| Statement  | Mean | Standard Deviation | Median | Mode |
|--|------|--------------------|--------|------|
| Familiar people can interpret her communication          | 4.41 | 0.79               | 5      | 5    |
| Unfamiliar people cannot interpret her communication     | 3.77 | 1.03               | 4      | 4    |
| Most people accurately estimate how much she understands | 2.17 | 1.08               | 2      | 2    |
| Most people underestimate the abilities of girls with RS | 4.30 | 1.19               | 5      | 5    |

In summary, the data presented in Table 7 indicate that for the communication effectiveness statements, the respondents perceived that familiar people are the ones who are most likely to accurately interpret the communication of girls with RS and that unfamiliar people have difficulty interpreting the communication behaviors of girls with RS. Respondents also perceived that most people do not accurately estimate how much a girls with RS can understand. The respondents also agreed that most people underestimate the abilities of girls with RS.

The distribution of the Likert Scale Scores for the remaining three statements is presented in Table 8. Respondents demonstrated strong agreement with the statement that they had confidence in their skills in working with girls with RS; for this question the mode was 5 (strongly agree) and the mean was 4.19. Forty-four percent of respondents chose this option, indicating strong agreement with this statement.

Respondents also agreed with the statement that there is information available to help them work with girls with RS; the mean for this statement was 3.84. The mode of 4 (agree) was chosen by 47% of respondents. The respondents also perceived that the communication skills in girls with RS have improved as a result of therapy, demonstrated by the mode of 4 (agree) for this question and the mean of 4.00. Forty-one percent of respondents agreed with this statement.

Table 8

Distribution of Likert scale Scores for Perceptions of Effectiveness of Therapy for RS, Respondent Skill Level in Working with RS, and Availability of Materials on RS

| Statement  | Mean | Standard Deviation | Median | Mode |
|--|------|--------------------|--------|------|
| Communication skills have improved with therapy    | 4.0  | 0.95               | 4      | 4    |
| Confidence in skills in working with girls with RS | 4.19 | 0.91               | 4      | 5    |
| Availability of information on working with RS     | 3.84 | 1.07               | 4      | 4    |

In summary, the data presented in Table 8 indicate that respondents perceive that their skills in working with girls with RS are strong, there is a great deal of information available publicly about RS, and that therapy is helpful for improving the communication skills of girls with RS.

### Nonparametric Statistical Analysis

The Chi-Square Test of Homogeneity was utilized to identify relationships between the independent and dependent variables using cross tabulations. Kendall's Tau-b was also calculated to determine the strength and direction of each association. Several significant relationships ( $p \leq .05$ ) were identified. The tables listing the Chi-Square  $p$  values and Kendall's Tau-b statistics for the significant associations are contained in Appendix F.

Analyses were conducted to determine if there were significant differences between the responses of the parent group and professional group to the 20 perception of communication statements (dependent variables). Calculations were made for all 20 statements. Five of the analyses met the criteria for statistical significance of  $p \leq .05$ . These five statements were: she communicates using spoken single words; she uses gestures to communicate; she understands at least 10 spoken words; familiar people can interpret her communication attempts; and most people underestimate the abilities of girls with RS. The data is presented in Table 9. There was no significant difference between perceptions of parents and professionals for the remaining 15 statements. For all of these associations, the results must be interpreted with caution, however, as there were numerous cells that had a count less than 5 responses per cell.

Table 9

Significant Differences Between Perceptions of Parents and Professionals in Response to Communication Perception Statements

| Communication Perception                                 | Pearson Chi-Square |    |                       | Kendall's Tau-b |             |
|--|--------------------|----|-----------------------|-----------------|-------------|
|  | Value              | Df | Asymp. Sig. (2-Sided) | Value           | Approx Sig. |
| She communicates using spoken single words               | 10.071             | 4  | .039(a)               | -.052           | .437        |
| She uses gestures to communicate                         | 13.028             | 4  | .011(a)               | -.088           | .183        |
| She understands at least 10 spoken words                 | 8.080              | 3  | .044(a)               | -.147           | .094        |
| Familiar people can interpret her communication attempts | 23.882             | 4  | .000(a)               | -.351           | .000(a)     |
| Most people underestimate the abilities of girls with RS | 16.720             | 4  | .002(a)               | -.268           | .003(a)     |

(a) – significant relationship

In summary, the data indicate that parents were significantly more likely than professionals to agree that a girl with RS used single words and gestures to communicate, and that the girl was also able to understand at least 10 spoken words. The relationship between role of respondent and the response to the statement was not particularly strong according to the Kendall's tau-b analysis, suggesting that other factors may play a role in the respondent's choice of answer.

A strong relationship was evident in the responses to the statement “familiar people can interpret her communication attempts” and the role of the respondent. The Kendall’s tau-b value was significant and suggested a stronger association between the variables than for the other responses. Specifically, parents were significantly more likely than professionals to agree or strongly agree that familiar people can interpret the communication attempts of girls with RS.

A relatively strong relationship was also evident between the role of the respondent and the response to the statement “most people underestimate the abilities of girls with RS”. The Kendall’s tau-b value was significant, which suggested a strong negative association between the variables. Here again, parents were more likely than professionals to strongly agree that most people underestimate the abilities of girls with RS.

In order to further explore the perceptions of the respondents, additional associations between responses to the perceptions were then studied. A significant association was demonstrated between history of speech use (question 10 in section 1 of the survey) and the response to the statement “she communicates using spoken single words”  $\chi^2(8, N=139) = 24.465, p = .002$ . A significant and strong negative relationship was identified between the variables  $\tau = -.336, p = .000$ . This indicated that a girl who achieved a level of oral speech

use at some time in her history was more likely to use single words at the time the survey was completed.

The relationships between use of spoken single words along with other communication modalities were explored. Significant relationships were found between responses to “she communicates using spoken single words” and 7 of the 19 other statements (Table 10). Strong, positive, and significant relationships were identified between responses to the statement regarding spoken single word use and girls’ use of sentences, use of sign, use of pictures/symbols/objects, use of gestures, use of body movements, and the ability to follow one-step commands. This suggests that respondents who agreed that the girl with RS used single words were also likely to agree that she uses multiple modalities to communicate, and that includes gestures, signs, and pictures, symbols or objects. The girl with RS who uses single words is also likely to use at least a few sentences to communicate and can follow one-step commands. As expected, a significant and strong relationship was identified between reported single word use and a significant cognitive impairment. This suggests that respondents who were more likely to agree that the girl with RS uses single words were also less likely to agree that she had a significant cognitive impairment.

Table 10

Significant relationships between responses to "She communicates using spoken single words" and responses to other communication perception questions

| Communication Perception                                     | Pearson Chi-Square |    |                       | Kendall's Tau-b |             |
|--|--------------------|----|-----------------------|-----------------|-------------|
|  | Value              | Df | Asymp. Sig. (2-sided) | Value           | Approx Sig. |
| She communicates using spoken sentences                      | 109.491            | 16 | .000                  | .592            | .000        |
| She communicates using sign(s)                               | 48.482             | 16 | .000                  | .334            | .000        |
| She uses pictures or symbols or other objects to communicate | 33.639             | 16 | .006                  | .282            | .000        |
| She uses gestures to communicate                             | 55.700             | 16 | .000                  | .324            | .000        |
| She use body movement to communicate                         | 26.791             | 16 | .044                  | .259            | .000        |
| She follows one-step commands                                | 26.713             | 16 | .045                  | .253            | .000        |
| She has a significant (severe) cognitive impairment          | 32.504             | 16 | .009                  | -.147           | .047        |

Note: all relationships significant

In the text responses that were given in section 3 of the survey, respondents were noted to describe apraxia as a factor in limiting an individual girl's ability to communicate and in increasing the time required for her to produce a response. Therefore, the association between apraxia (motor planning



abilities) and cognition and the perceptions of others was explored by analyzing 3 sets of related variables. All three of these associations were found to be significant (Table 11). Respondents who agreed with the statement that apraxia interfered with communication were also significantly less likely to agree that the girl with RS had a significant cognitive impairment. Respondents who agreed with the statement that apraxia interfered with communication were also significantly less likely to agree that people were accurately able to estimate how much a girl with RS can understand. Respondents who agreed that there is a relationship between the influence of apraxia on communication, also were significantly likely to agree that other people underestimate the abilities of girls with RS. The relationship between this last set of responses was also found to be relatively strong based,  $\tau = .274$ ,  $p = .000$ , compared to the previous responses.

Table 11

Significant relationships between responses to “Apraxia (motor planning problem) limits her ability to communicate with others” and three communication perception questions

| Communication Perception   | Pearson Chi-Square |    |                       | Kendall's Tau-b |              |
|--|--------------------|----|-----------------------|-----------------|--------------|
|  | Value              | Df | Asymp. Sig. (2-sided) | Value           | Approx. Sig. |
| She has a significant (severe) cognitive impairment                | 31.658             | 16 | .011(a)               | -.125           | .089         |
| Most people accurately estimate how much she knows and understands | 29.811             | 16 | .019(a)               | -.108           | .172         |
| Most people underestimate the abilities of girls with RS           | 39.993             | 16 | .001(a)               | .274            | .000 (a)     |

(a) – significant relationship

### Statistical Analysis of Group Differences

The Mann-Whitney U test was used to determine if significant differences existed between the responses from the parents and professionals. Mean ranks and summed ranks were calculated on the responses to the twenty Likert statements. The differences in mean rank between the groups were significant for only 3 of the 20 statements (Table 12).

Table 12  
Significant Mann-Whitney U calculations

| Statements   | Mean Ranks |               | Asymp. Sig. (2-tailed) |
|--|------------|---------------|------------------------|
|  | Parents    | Professionals |                        |
| She communicates using vocalizations                     | 74.72      | 53.76         | .012                   |
| Familiar people can interpret her communication attempts | 77.11      | 42.66         | .000                   |
| Most people underestimate the abilities of girls with RS | 75.55      | 49.88         | .001                   |

The Mann-Whitney U test results indicate that though there were differences in the parents and professionals in how they responded to the 20 Likert scale statements, the group differences were only significant for the responses to the 3 statements that appear in Table 12. Parents were significantly more likely than professionals to indicate agreement with the following statements: a girl with RS uses vocalizations to communicate, familiar people can interpret her communication attempts, and most people underestimate the abilities of girls with RS.

## Responses to Open-Ended Question Regarding Communication Skills

Ninety-one respondents (65%) wrote text messages in section 3 of the survey, in response to the statement: "Please add any additional information regarding the girl with RS and her communication skills, such as communication strategies, words/phrases used, any augmentative systems, or anything unique or special about her communication skills". The responses were analyzed by identifying occurrences of keywords or phrases which were part of the research questions or hypotheses that were posed at the start of the study.

The primary topic in the text messages was a description of an individual girl's communication system. Respondents used this section to describe a girl's evolution of communication skills or to describe her current system and each individual modality. As indicated by the descriptive statistical analysis of the data, respondents reported that girls with RS used multiple communication modalities.

At the start of the study, three research questions were posed regarding the types of information that would be shared in the text messages. Messages that were related to these three questions are presented in Appendix G under three themes: 1) skills of girls with RS are underestimated; 2) inconsistency of responses is a barrier to education or communication; and 3) strategies used to enhance communication. Thirty-nine messages (or sections of messages) are listed in Appendix H. Twelve of these messages dealt specifically with issues

regarding underestimation of skills of girls with RS. They confirm the results obtained from the analysis of the responses to the Likert scale statements. Twelve messages described inconsistent skills evidenced by girls with RS. As was revealed in the earlier statistical analysis, respondents ascribe many of the difficulties experienced in RS to problems from apraxia. Strategies that are used to successfully enhance communication interactions are described in fifteen text messages. As was evident from the earlier statistical analysis, respondents report that using multiple modes of communication (eye gaze, access to AAC systems, vocalizations, and body movements) allow girls with RS to express a multitude of communicative intentions and engage in successful social interactions.

## Chapter V

### DISCUSSION

#### Significant Findings

The purpose of this study was to gather data on the communication skills of girls with Rett syndrome (RS) as perceived by people most familiar with this population, their parents and professionals (teachers and speech-language pathologists [SLPs]). Data was gathered through use of an online survey that was publicized to members of the Rett Syndrome community around the world.

A variety of relationships were explored regarding communication abilities in RS, including relationships between history of speech use and current communication skills, use of communication modalities and assessments of communication partners, methods used to access communication systems and comprehension skills, and the relationship between current speech use and assessments of other skills by participants. Similarities and differences were analyzed between the participants as defined by role, whether the respondent was a parent or professional (teacher or SLP). Respondents were asked to describe the current communication abilities of a girl with RS, and the information

they reported was analyzed in regard to the potential for underestimation of ability, the possibility that inconsistent behaviors are a potential barrier to education or communication, and the types of strategies that are being utilized to enhance communication in girls with RS.

Five hypotheses were posed at the start of the study. The majority of girls with RS were expected to use more than one communication modality (e.g., eye gaze, vocalization, speech, gesture, body movements, picture/symbol system) to communicate. Eye gaze was predicted to be the most frequently used modality for communication by the girls with RS. All participants were expected to agree that the cognitive abilities of girls with RS are underestimated. Parents were predicted to report that the girls' comprehension abilities are greater relative to the reports by the professionals. The majority of participants were predicted to agree that motor planning difficulties impair the ability of girls with RS to communicate. The results indicated that all hypotheses were supported.

A total of 141 respondents completed the online survey during the allotted time period, May 1, 2005 – June 30, 2005. The sample consisted of 116 parents and 25 professionals. The respondents resided in at least 9 countries, with the majority living in the United States. They reported on the communication of girls with RS across the lifespan. The majority of the girls with RS are in some type of educational program at the current time. Most have benefited from a variety of rehabilitation services, including speech, physical and occupational therapy. Half

of the girls have received an augmentative communication evaluation. Fully one-third of the girls produce responses after a delay of between 6-10 seconds. Fifty-seven percent of the girls are reported to produce responses after delays of longer than 10 seconds. Of those girls who experience the longer delays, fully 20% produce responses after 30 seconds or more.

Nineteen percent of girls are using some amount of oral speech at the current time, with the majority using approximately 1- 10 single words. As predicted, most girls are using more than one communication modality at the current time, with the mean being 3.25 and the mode being 4 modalities. The most frequently used communication modality, as predicted, was eye gaze, which is used by 79% of the girls with RS. Respondents strongly agreed that this was a modality used most frequently by girls with RS. Interestingly, there was little variability around the mean for this response, indicating strong agreement across both groups of participants (parents and professionals). Body movement and pictures or symbol boards were the next most frequently used modalities for communication. These findings have implications for both augmentative communication evaluations and for therapeutic programming. Since eye gaze appears to be an effective and consistently utilized communication modality, therapists and parents should explore an individual girl's use of this modality and program intervention accordingly.



Parents and professionals alike strongly disagreed that signs were used by girls as a communication modality. This finding is not surprising given the strong agreement among respondents that apraxia limited the ability of girls to communicate. Apraxia, which is a motor planning disorder, is believed to influence the motoric abilities of girls with RS. The degree to which individual girls are affected is unclear, and there are no reliable tests that can measure the presence/absence or degree of apraxia in this population. The existence of an apraxia in an individual girl is typically determined by experienced professionals who work with her over a period of time, and evaluate her responses to stimuli under multiple conditions. If an individual girl has apraxia, her ability to utilize signs (as in sign language) would be limited because of motoric constraints. Likewise, her ability to access picture or symbol boards using her hands would be affected.

A significant relationship was also identified between apraxia and cognitive impairment in RS as predicted. Respondents perceived that apraxia influences how the cognitive potential of girls with RS can be estimated. Further, respondents indicated that people were more likely to underestimate the abilities of girls with RS particularly when apraxia was observed. Respondents strongly agreed that girls with RS present with a significant cognitive impairment. This finding is consistent with previous literature (Cass et al., 2003). What hasn't been previously reported is the perception that cognitive impairment and apraxia interact to affect communication and that this has strong implications for

assessment and intervention. This is clearly an area that deserves further study and analysis.

A strong and significant relationship was also identified between history of speech use and current use of speech for communication. This indicates that history of using speech prior to regression is associated with later use of oral speech for communication. However, the anecdotal reports provided in section 3 of the survey indicated that most girls who are using some type of verbal speech have a limited vocabulary of between 1-10 single words. This is another area that bears further investigation. The types of words and the communicative functions that are conveyed by oral speech could be explored in a clinical study of girls with RS. It has been reported that girls who are verbal are more likely to speak some words during periods of extreme excitement, agitation or when highly motivated (Zappella et al., 1998). Perhaps high levels of agitation affect the central nervous system in some way, overcoming the influence of apraxia.

There were surprisingly few differences between the professionals and parents in their responses to the survey questions. We predicted that parents would report that the girls' comprehension abilities were higher than the reports of the professionals. However, only 5 of the 20 statements results in significant differences between the perceptions of parents and professionals.

Parents were more likely than the professionals to agree that a girl with RS uses single words to communicate, uses gestures to communicate, and is able to understand at least 10 single words. Though the differences for these three statements were significant based on role of the subject, the associations between role of subject and each of the three statements were weak. This findings suggests that for these three specific skill areas (speech, gestures, comprehension of single words), parents are more likely to rate their daughter's skills as higher than the professionals will rate the skills. This finding is not surprising given reports in the literature that question the ability of parents to accurately describe their daughter's abilities, and a reported tendency by parents to overestimate the skill level of their daughter (Koppenhaver et al., 2001).

A significant and fairly strong association was identified between the role of the participant (parent or professional) and the response to the statement "Familiar people can interpret the communication skills of girls with RS". Parents were more likely than professionals to strongly agree with this statement though there was fairly little variability in responses overall ( $SD = 0.79$ ) and the median and mode were both 5 (strongly agree) for this statement. Professionals were more likely to choose "agree" than parents. All respondents, regardless of role, were unlikely to choose an alternative response. Of the 141 total responses for this statement, only 9 respondents chose "undecided or unsure", 2 chose "disagree" and 2 chose "strongly disagree". This suggests that respondents, both parents and professionals, believe that familiar people can interpret the

communication skills of girls with RS. This has implications for future research regarding the communication skills of individual girls with RS. A study could be designed explore the communicative abilities of a group of girls with RS, and use interactions with parents and familiar professionals as contexts in which assessments of communicative interactions are made. In these dynamic contexts, the role of the communication partner, and the communicative effort expended by these partners could be analyzed for each dyad. Outcomes could be used to design successful communicative interventions for girls with RS.

The final difference that was identified between the groups on responses to the Likert scale statements was in regard to the statement: "Most people underestimate the abilities of girls with RS". There was a fairly strong and significant association between role of the subject and the response to this statement. The data indicate that although the respondents overall tended to strongly agree with this statement (median and mode were both 5), the parents were significantly more likely than the professionals to choose 5 (strongly agree) as a response. Professionals were more likely to choose 4 (agree) than any other response choice.

It was actually surprising to the researcher that the ratings of the parents and professionals did not differ significantly on more Likert scale statements. Results of the Mann Whitney U tests on responses to the Likert scale questions indicated that differences in the two groups were not significant on seventeen of

the twenty statements. Group differences were evident in responses to the statements regarding use of vocalizations to communicate, perceptions of ability of familiar people to interpret communication attempts by girls with RS, and tendency of most people to underestimate the abilities of girls with RS. Parents used a higher ranking than professionals on each of these statements, indicating a tendency to strongly agree with the statements. Most professionals tended to agree with the statements. Though the results of the Mann Whitney U test indicate that these two groups have some inherent differences, their overall ratings were fairly similar for the majority of responses.

### Limitations

One of the limitations of the study was the difference in the size of the two groups of respondents. A total of 141 respondents submitted completed surveys; of those, 116 were parents and 25 were professionals. It was therefore less than ideal to calculate group differences when the sizes of the two groups were so disparate. Additionally, the professional group was comprised of 14 SLPs and 11 teachers. There may be differences among these two subgroups of professionals that further influenced the data.

The respondents that completed the survey represented a convenience sample of volunteers who learned about the survey through an email or a flier distributed at an international conference or their child's (or student's) school. It is possible that the group that responded to the survey is not representative of

other persons in the Rett syndrome community who do not have access to the methods of solicitation for this project. Since the survey was completed anonymously and on-line, the veracity of the responses cannot be verified. The use of a password was added to make it less likely that the same person would answer the survey more than one time. However, this could have been circumvented by the use of an alternate password.

Another limitation was in regard to the construction of the Likert scale questions. A five-point scale was utilized in the survey and ordinal data was collected. The researcher was unable to determine if the categories (strongly agree, agree, etc.) have equal intervals between them. It is actually highly unlikely that the intervals between the categories are all equal.

Lastly, the inability to collect data on the stage of RS was a clear limitation. It was likely that most parents could reliably report which stage of RS a particular girl was in, but the results of the pilot study revealed that professionals were unable to provide that data. It would have been helpful to be able to analyze the relationship between stage of RS and level of communication skill as part of this study. Perhaps that can be explored in future research.

## Chapter VI

### SUMMARY AND CONCLUSIONS

The purpose of this study was to expand the body of literature on communication skills in girls with Rett Syndrome (RS). The results of this study indicate that girls with RS are perceived by familiar communication partners as capable of communication. Girls with RS are reported to use multiple modalities to communicate, including eye gaze, body movements, and augmentative communication systems that contain pictures or symbols. Respondents indicate that apraxia, a motor planning impairment, has a strong influence on a girl with RS' ability to communicate with others. Apraxia is also perceived to influence consistency and speed of motor response, and to influence how well others can judge the cognitive and comprehension skills of a girl with RS. This study should provide direction for future research on effective ways to assess communication abilities and potential in this population, and how to design interventions that maximally utilize the skills of this population.

Eye gaze was reported to be the most commonly used communication modality in this study. Respondents indicated that girls with RS may use eye gaze in many different ways. A girl may use eye gaze informally without any prior

training, as in looking intensely at a desired object. In this circumstance, the burden of communication is on the partner, who must recognize that the eye gaze pattern is intentional and that the girl with RS is using eye gaze to make a request. This behavior must be verified over multiple trials and with different partners. Those persons who interact with the girl with RS must then be trained to identify those instances in which the girl is using her eye gaze intentionally. This communication modality can then be further developed to see if eye blinks (for a yes/no response) can be trained, or if a girl can be trained to use mutual eye gaze behavior to first look at a desired object and then look at the partner to acknowledge the request. We have much to learn about mutual eye gaze behavior in RS. There is some evidence that girls with RS do not use mutual eye gaze for communicative purposes (Cass et al., 2003). This is an area in need of further clinical exploration for assessment and intervention (G. Woodyatt, personal communication, March 31, 2005).

There are a wide variety of eye gaze systems that can be used for augmentative communication. There are low-tech options that use direct eye gaze at 1-2 objects or pictures of objects. More complex, non-electronic eye gaze systems can be established that use 1- or 2-step processes, depending upon the field of choices that are available. There are fairly complex, electronic eye gaze systems that can use digital technology to recognize the cornea of a user. It is important that clinicians who perform augmentative communication



evaluations become cognizant of the strong eye gaze behaviors of girls with RS and use this modality in planning interventions and communication systems.

The data from this study revealed that girls were using multiple modalities to communicate and that the mean number was 3+ modalities per girl. This finding also has implications for communication assessment and intervention planning. Clinicians should evaluate all modalities and consider the influence of apraxia, delayed response time, and inconsistency in responding when planning interventions. It became clear from examining the data and reading the text responses that girls may use different modalities based on multiple factors. Seizure activity and level of awareness have been reported as two factors which can influence quality of responsiveness in RS. By providing a girl with RS multiple modalities to communicate, and by training communication partners to recognize level of alertness and types of responses, the communicative and educational potential of these girls may be better realized. Outcome data on effectiveness of types of interventions in this population is clearly needed based on the findings of this study.

This study demonstrated that the perceptions of parents and professionals with regard to the communication skills of girls with RS are fairly similar. What is also clear is that familiarity with an individual girl is important. As is common when interacting with any individual with a severe disability, the communication partner must use inference and intuition in order to ascribe meaning to the

person's behaviors. This does not mean that the communication partner is overestimating competence on the part of the disabled individual. Rather, there is an assumption that competent behavior is possible, and then significant work on the part of the communication partner to accurately interpret the signals. This study has demonstrated that girls with RS are communicating to their parents, their teachers and the speech-language pathologists who work with them. All of these individuals are working hard to untangle the meanings conveyed by the girls with RS and to accurately interpret their behaviors. There is much work to be done to develop clinical processes for accurate assessments that can be utilized in the presence of severe disability. We can use the perceptions of these familiar partners, combined with their rich descriptions of the behaviors they observe, to develop objective measures of skill level and outcomes of the intervention we provide to girls with RS.

The girls with RS have much to say to us: we have to know how to listen and "read" their skills, how best to teach them, and how to help them say even more.

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## Appendix A

### Diagnostic Criteria for Rett Syndrome

#### Necessary Criteria

1. apparently normal prenatal and perinatal history
2. psychomotor development largely normal through the first six months or may be delayed from birth
3. normal head circumference at birth
4. postnatal deceleration of head growth in the majority
5. loss of achieved purposeful hand skill between ages  $\frac{1}{2}$  - 2  $\frac{1}{2}$  years
6. stereotypic hand movements such as hand wringing/squeezing, clapping/tapping, mouthing and washing/rubbing automatisms
7. emerging social withdrawal, communication dysfunction, loss of learned words, and cognitive impairment
8. impaired (dyspraxic) or failing locomotion

#### Supportive Criteria

1. awake disturbances of breathing (hyperventilation, breath holding, forced expulsion of air or saliva, air swallowing)
2. bruxism
3. impaired sleep pattern from early infancy
4. abnormal muscle tone successively associated with muscle wasting and dystonia
5. peripheral vasomotor disturbances
6. scoliosis/kyphosis progressing through childhood
7. growth retardation
8. hypotrophic small and cold feet; small, thin hands

#### Exclusion Criteria

1. organomegaly or other signs of storage disease
2. retinopathy, optic atrophy, or cataract
3. evidence of perinatal or postnatal brain damage
4. existence of identifiable metabolic or other progressive neurological disorder
5. acquired neurological disorder resulting from severe infections or head trauma

## Revised delineation of variant phenotypes

### Inclusion criteria

1. meet at least 3 of 6 main criteria
2. meet at least 5 of 11 supportive criteria

### Six main criteria

1. absence or reduction of hand skills
2. reduction or loss of babble speech
3. monotonous pattern of hand stereotypies
4. reduction or loss of communication skills
5. deceleration of head growth from first years of life
6. RS disease profile; a regression stage followed by a recovery of interaction contrasting with slow neuromotor regression

### Eleven supportive criteria

1. breathing irregularities
2. bloating/air swallowing
3. teeth grinding, harsh sounding type
4. abnormal locomotion
5. scoliosis/kyphosis
6. lower limb amyotrophy
7. cold, purplish feet, usually growth impaired
8. sleep disturbances including night screaming outbursts
9. laughing/screaming spells
10. diminished response to pain
11. intense eye contact/eye pointing

(International Rett Syndrome Association, 2002)



## SETON HALL UNIVERSITY

1 8 5 6

March 30, 2005

Theresa Bartolotta  
5 Coronet Avenue  
Lincroft, NJ 07738

Dear Professor Bartolotta,

The Seton Hall University Institutional Review Board has reviewed and approved as submitted under exempt review your research proposal entitled "Communication Skills in Girls with Rett Syndrome: Perceptions of Parents, Speech-language Pathologist and Teachers". The IRB reserves the right to recall the proposal at any time for full review.

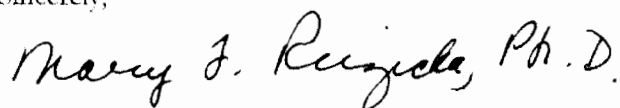
Enclosed for your records is the signed Request for Approval form.

The Institutional Review Board approval of your research is valid for a one-year period from the date of this letter. During this time, any changes to the research protocol must be reviewed and approved by the IRB prior to their implementation.

According to federal regulations, continuing review of already approved research is mandated to take place at least 12 months after this initial approval. You will receive communication from the IRB Office for this several months before the anniversary date of your initial approval.

Thank you for your cooperation.

Sincerely,



Mary F. Ruzicka, Ph.D.  
Professor  
Director, Institutional Review Board

cc Genevieve Pinto-Zipp, Ph.D.

Office of Institutional Review Board  
Presidents Hall  
Tel: 973 313.6314 • Fax: 973 275.2978  
400 South Orange Avenue • South Orange, New Jersey 07079-2641

## Appendix C

### **Subject : Invitation to Participate in Research about Rett Syndrome**

Dear Member of the Rettnet:

A research study investigating the communication behaviors of girls with Rett Syndrome is being conducted. I am seeking parents of girls with Rett Syndrome and teachers and speech-language pathologists who work with a girl with Rett Syndrome. Participants will complete an on-line survey about the communication skills of the girl with Rett Syndrome. The survey will take approximately 15 minutes to complete.

There will be no cost involved in participating in this research. All responses will be anonymous and confidential.

To take the on-line survey, log on to:

**<http://tltc.shu.edu:80/servlets/asset.AssetSurvey?surveyid=791>**

If you have any questions about your participation in this project, please contact Theresa E Bartolotta, principal investigator, at the location below:

Theresa E Bartolotta  
Assistant Professor  
Department of Communication Disorders & Deafness  
Kean University  
Union, New Jersey  
Phone: 908-737-5408  
Email: [tbartolo@kean.edu](mailto:tbartolo@kean.edu)

This research has been approved by the Institutional Review Board of Seton Hall University.

## Appendix D

### **Invitation to Participate in Research about Rett Syndrome**

A research study investigating the communication behaviors of girls with Rett Syndrome is being conducted. I am seeking parents of girls with Rett Syndrome and teachers and speech-language pathologists who work with a girl with Rett Syndrome. Participants will complete an on-line survey about the communication skills of the girl with Rett Syndrome. The survey will take approximately 15 minutes to complete.

There will be no cost involved in participating in this research. All responses will be anonymous and confidential.

To take the on-line survey, log on to:

**<http://tltc.shu.edu:80/servlets/asset.AssetSurvey?surveyid=791>**

If you have any questions about your participation in this project, please contact Theresa E Bartolotta, principal investigator, at the location below:

Theresa E Bartolotta  
Assistant Professor  
Department of Communication Disorders & Deafness  
Kean University  
Union, New Jersey  
Phone: 908-737-5408  
Email: [tbartolo@kean.edu](mailto:tbartolo@kean.edu)

This research has been approved by the Institutional Review Board of Seton Hall University.

## Appendix E

**COMMUNICATION SKILLS IN GIRLS WITH RETT SYNDROME**

*This survey will measure the perceptions of the communication skills of girls with Rett syndrome by parents, speech-language pathologists and teachers.*

**I. Background Information****1. For the person completing the survey – are you a**

- ☐ *Parent or Caregiver of a girl with Rett Syndrome (RS)*
- ☐ *Speech-language pathologist currently working with a girl with RS*
- ☐ *Teacher of a girl with RS*

**2. Where do you live?**

- ☐ *Northeastern United States*
- ☐ *Southeastern United States*
- ☐ *Northwestern United States*
- ☐ *Southwestern United States*
- ☐ *Australia*
- ☐ *Brazil*
- ☐ *England*
- ☐ *Germany*
- ☐ *Ireland*
- ☐ *Italy*
- ☐ *Mexico*
- ☐ *New Zealand*
- ☐ *Portugal*
- ☐ *Scotland*
- ☐ *South Africa*
- ☐ *Spain*
- ☐ *Turkey*
- ☐ *Other*

**Please complete the following questions relative to your daughter with RS or a girl you are currently working with who has RS.**

**3. Age of girl with RS**

- ☐ 0-3 years
- ☐ 4-7 years
- ☐ 8-13 years
- ☐ 13-20 years
- ☐ 21 years+

**4. Does she use any of the following modalities to communicate? Please check all that apply.**

- ☐ Signs
- ☐ Verbal Speech
- ☐ Pictures/Symbols/Objects (with a switch or communication or gaze board)
- ☐ Eye contact/eye gaze/eye pointing/eye blinking
- ☐ Body movement (kicking, walking towards something)
- ☐ Gestures (pointing, head shaking, head turning)
- ☐ Voice output device (electronic device that "speaks")
- ☐ Other

**5. If she uses pictures, symbols or objects to communicate, how does she access them?**

- ☐ Touch (with hand or fingers)
- ☐ Eye pointing or eye gaze
- ☐ Head pointer (stick or laser)
- ☐ Other
- ☐ Not applicable

**6. On average, how long does it take for her to produce a response?**

- ☐ 0-5 seconds
- ☐ 6-10 seconds
- ☐ 11-20 seconds
- ☐ 21-30 seconds
- ☐ More than 30 seconds

**7. Is she currently receiving, or has she ever received, the following services (check all that apply):**

- ☐ *Speech therapy*
- ☐ *Occupational therapy*
- ☐ *Physical therapy*
- ☐ *Music therapy*
- ☐ *Augmentative Communication evaluation*
- ☐ *Other*

**8. Is she currently enrolled in an educational program?**

- ☐ *Yes*
- ☐ *No*

**9. If yes, please choose the setting that best applies:**

- ☐ *Special school for children with disabilities*
- ☐ *Special class in a regular school*
- ☐ *Regular class with typical children*
- ☐ *Home instruction*
- ☐ *Adult day program*
- ☐ *Not applicable*

**10. Did she ever speak or use words?**

- ☐ *Yes (go on to the next question)*
- ☐ *No, she never used words in the past and does not speak now (go to Section II Communication Skills, question #16)*

**11. Has she lost her speech or ever had a regression in speech?**

- ☐ *Yes*
- ☐ *No*
- ☐ *Don't know*

**12. How old was she when she lost her speech or had a regression in speech?**

- ☐ *Less than 1 year of age*
- ☐ *Between 1 and 2 years of age*
- ☐ *Between 2 and 3 years of age*
- ☐ *Older than 3 years*

**13. How much language or speech did she have before she lost her speech?**

- ☐ *Less than 5 single words*  
☐ *10 to 50 single words*  
☐ *She used short phrases*  
☐ *She used sentences*  
☐ *She never spoke*

**14. Does she speak now?**

- ☐ *Yes*  
☐ *No*

**15. What words or phrases does she say? (Please list them)**

---



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## **II. Communication Skills**

**Please indicate your level of agreement with the following statements regarding the communication abilities of the girl with RS.**

| <b>16.</b>  | <i>Strongly Agree</i> | <i>Agree</i> | <i>Unsure or Undecided</i> | <i>Disagree</i> | <i>Strongly Disagree</i> |
|---|-----------------------|--------------|----------------------------|-----------------|--------------------------|
| <i>She communicates using spoken single words</i>                   |                       |              |                            |                 |                          |
| <i>She communicates using spoken sentences</i>                      |                       |              |                            |                 |                          |
| <i>She communicates using vocalizations (e.g. sounds, yelling)</i>  |                       |              |                            |                 |                          |
| <i>She communicates using signs</i>                                 |                       |              |                            |                 |                          |
| <i>She uses pictures of symbols or other objects to communicate</i> |                       |              |                            |                 |                          |

**Please indicate your level of agreement with the following statements regarding the communication abilities of the girl with RS.**

| 17.  | <i>Strongly Agree</i> | <i>Agree</i> | <i>Unsure or Undecided</i> | <i>Disagree</i> | <i>Strongly Disagree</i> |
|--|-----------------------|--------------|----------------------------|-----------------|--------------------------|
| <i>She uses gestures (e.g. pointing, head shake) to communicate</i>                        |                       |              |                            |                 |                          |
| <i>She uses eye gaze (e.g. eye gaze, eye contact, eye point, eye blink) to communicate</i> |                       |              |                            |                 |                          |
| <i>She uses body movement (e.g. kicking, moving towards an object) to communicate</i>      |                       |              |                            |                 |                          |
| <i>She can read one or more single words</i>   |                       |              |                            |                 |                          |
| <i>She understands at least 10 spoken words (e.g. single words, names of people)</i>       |                       |              |                            |                 |                          |



**Please indicate your level of agreement with the following statements regarding the communication abilities of the girl with RS.**

| <b>18.</b>  | <i>Strongly Agree</i> | <i>Agree</i> | <i>Unsure or Undecided</i> | <i>Disagree</i> | <i>Strongly Disagree</i> |
|---|-----------------------|--------------|----------------------------|-----------------|--------------------------|
| <i>She follows one-step commands (e.g. go into the bathroom)</i>                      |                       |              |                            |                 |                          |
| <i>She has a significant (severe) cognitive impairment</i>                            |                       |              |                            |                 |                          |
| <i>Apraxia (motor planning problem) limits her ability to communicate with others</i> |                       |              |                            |                 |                          |
| <i>Familiar people can interpret her communication attempts</i>                       |                       |              |                            |                 |                          |
| <i>Unfamiliar people cannot understand her communication attempts</i>                 |                       |              |                            |                 |                          |

**Please indicate your level of agreement with the following statements regarding the communication abilities of the girl with RS.**

| <b>19.</b>   | <i>Strongly Agree</i> | <i>Agree</i> | <i>Unsure or Undecided</i> | <i>Disagree</i> | <i>Strongly Disagree</i> |
|--|-----------------------|--------------|----------------------------|-----------------|--------------------------|
| <i>Most people accurately estimate how much she knows and understands</i>  |                       |              |                            |                 |                          |
| <i>Her communication skills have improved with training and/or therapy</i> |                       |              |                            |                 |                          |
| <i>I am confident of my skills in working with this girl with RS</i>       |                       |              |                            |                 |                          |
| <i>I was able to find information on how to work with girls with RS</i>    |                       |              |                            |                 |                          |
| <i>Most people underestimate the abilities of girls with RS</i>            |                       |              |                            |                 |                          |

### **III. Additional Information**

Please add any additional information regarding the girl with Rett Syndrome and her communication skills, such as communication strategies, words/phrases used, any augmentative systems, or anything unique or special about her communication skills.

## Appendix F

### Significant Chi Square Relationships

Significant relationship between role of respondent and response to "She communicates using spoken single words"

#### Chi-Square Tests

|                                 | Value     | df | Asymp. Sig.<br>(2-sided) |
|---------------------------------|-----------|----|--------------------------|
| Pearson Chi-Square              | 10.071(a) | 4  | .039                     |
| Likelihood Ratio                | 12.473    | 4  | .014                     |
| Linear-by-Linear<br>Association | 2.450     | 1  | .118                     |
| N of Valid Cases                | 141       |    |                          |

a. 4 cells (40.0%) have expected count less than 5. The minimum expected count is .89.

#### Symmetric Measures

|                                       | Value | Asymp.<br>Std.<br>Error(a) | Approx.<br>T(b) | Approx. Sig. |
|---------------------------------------|-------|----------------------------|-----------------|--------------|
| Ordinal by Ordinal    Kendall's tau-b | -.052 | .066                       | -.777           | .437         |
| N of Valid Cases                      | 141   |                            |                 |              |

a. Not assuming the null hypothesis.

b. Using the asymptotic standard error assuming the null hypothesis.

Significant relationship between role of participant and response to "She uses gestures to communicate"

#### Chi-Square Tests

|                                 | Value     | df | Asymp. Sig.<br>(2-sided) |
|---------------------------------|-----------|----|--------------------------|
| Pearson Chi-Square              | 13.028(a) | 4  | .011                     |
| Likelihood Ratio                | 13.519    | 4  | .009                     |
| Linear-by-Linear<br>Association | 1.057     | 1  | .304                     |
| N of Valid Cases                | 141       |    |                          |

a. 2 cells (20.0%) have expected count less than 5. The minimum expected count is 2.48.

#### Symmetric Measures

|                                       | Value | Asymp.<br>Std.<br>Error(a) | Approx.<br>T(b) | Approx. Sig. |
|---------------------------------------|-------|----------------------------|-----------------|--------------|
| Ordinal by Ordinal    Kendall's tau-b | -.088 | .065                       | -1.330          | .183         |
| N of Valid Cases                      | 141   |                            |                 |              |

a. Not assuming the null hypothesis.

b. Using the asymptotic standard error assuming the null hypothesis.

Significant relationship between role of respondent and response to "She understands at least 10 spoken words"

#### Chi-Square Tests

|                                 | Value    | df | Asymp. Sig.<br>(2-sided) |
|---------------------------------|----------|----|--------------------------|
| Pearson Chi-Square              | 8.080(a) | 3  | .044                     |
| Likelihood Ratio                | 7.322    | 3  | .062                     |
| Linear-by-Linear<br>Association | .785     | 1  | .376                     |
| N of Valid Cases                | 141      |    |                          |

a. 4 cells (50.0%) have expected count less than 5. The minimum expected count is .35.

#### Symmetric Measures

|                                    | Value | Asymp.<br>Std.<br>Error(a) | Approx.<br>T(b) | Approx. Sig. |
|------------------------------------|-------|----------------------------|-----------------|--------------|
| Ordinal by Ordinal Kendall's tau-b | -.147 | .086                       | -1.673          | .094         |
| N of Valid Cases                   | 141   |                            |                 |              |

a. Not assuming the null hypothesis.

b. Using the asymptotic standard error assuming the null hypothesis.

Significant relationship between role of respondent and response to "Familiar people can interpret her communication attempts"

#### Chi-Square Tests

|                              | Value     | df | Asymp. Sig. (2-sided) |
|------------------------------|-----------|----|-----------------------|
| Pearson Chi-Square           | 23.882(a) | 4  | .000                  |
| Likelihood Ratio             | 21.723    | 4  | .000                  |
| Linear-by-Linear Association | 20.477    | 1  | .000                  |
| N of Valid Cases             | 141       |    |                       |

a. 5 cells (50.0%) have expected count less than 5. The minimum expected count is .35.

#### Symmetric Measures

|                                    | Value | Asymp. Std. Error(a) | Approx. T(b) | Approx. Sig. |
|------------------------------------|-------|----------------------|--------------|--------------|
| Ordinal by Ordinal Kendall's tau-b | -.351 | .075                 | -3.980       | .000         |
| N of Valid Cases                   | 141   |                      |              |              |

a. Not assuming the null hypothesis.

b. Using the asymptotic standard error assuming the null hypothesis.

Significant relationship between role of respondent and response to "Most people underestimate the abilities of girls with RS"

#### Chi-Square Tests

|                                 | Value     | df | Asymp. Sig.<br>(2-sided) |
|---------------------------------|-----------|----|--------------------------|
| Pearson Chi-Square              | 16.720(a) | 4  | .002                     |
| Likelihood Ratio                | 14.053    | 4  | .007                     |
| Linear-by-Linear<br>Association | 8.224     | 1  | .004                     |
| N of Valid Cases                | 141       |    |                          |

a. 5 cells (50.0%) have expected count less than 5. The minimum expected count is 1.06.

#### Symmetric Measures

|                                    | Value | Asymp.<br>Std.<br>Error(a) | Approx.<br>T(b) | Approx. Sig. |
|------------------------------------|-------|----------------------------|-----------------|--------------|
| Ordinal by Ordinal Kendall's tau-b | -.268 | .083                       | -2.988          | .003         |
| N of Valid Cases                   | 141   |                            |                 |              |

a. Not assuming the null hypothesis.

b. Using the asymptotic standard error assuming the null hypothesis.

Significant relationship between history of speech use and response to "She communicates using spoken single words"

#### Chi-Square Tests

|                              | Value     | df | Asymp. Sig. (2-sided) |
|------------------------------|-----------|----|-----------------------|
| Pearson Chi-Square           | 24.465(a) | 8  | .002                  |
| Likelihood Ratio             | 33.621    | 8  | .000                  |
| Linear-by-Linear Association | 17.132    | 1  | .000                  |
| N of Valid Cases             | 139       |    |                       |

a. 9 cells (60.0%) have expected count less than 5. The minimum expected count is .25.

#### Symmetric Measures

|                                    | Value | Asymp. Std. Error(a) | Approx. T(b) | Approx. Sig. |
|------------------------------------|-------|----------------------|--------------|--------------|
| Ordinal by Ordinal Kendall's tau-b | -.336 | .056                 | -5.439       | .000         |
| N of Valid Cases                   | 139   |                      |              |              |

a. Not assuming the null hypothesis.

b. Using the asymptotic standard error assuming the null hypothesis.

Note: 2 of the 141 cases did not answer the question "Did she ever speak or use words?", therefore this data is based on 139 responses.



Significant relationship between responses to “Apraxia limits her ability to communicate with others” and “Most people underestimate the abilities of girls with RS”

#### Chi-Square Tests

|                              | Value     | df | Asymp. Sig. (2-sided) |
|------------------------------|-----------|----|-----------------------|
| Pearson Chi-Square           | 39.993(a) | 16 | .001                  |
| Likelihood Ratio             | 41.204    | 16 | .001                  |
| Linear-by-Linear Association | 3.292     | 1  | .070                  |
| N of Valid Cases             | 141       |    |                       |

a. 19 cells (76.0%) have expected count less than 5. The minimum expected count is .13.

#### Symmetric Measures

|                    |                 | Value | Asymp. Std. Error(a) | Approx. T(b) | Approx. Sig. |
|--------------------|-----------------|-------|----------------------|--------------|--------------|
| Ordinal by Ordinal | Kendall's tau-b | .274  | .076                 | 3.599        | .000         |
| N of Valid Cases   |                 | 141   |                      |              |              |

a. Not assuming the null hypothesis.

b. Using the asymptotic standard error assuming the null hypothesis.

Significant relationship between responses to "She has a significant (severe) cognitive impairment" and "Apraxia (motor planning problem) limits her ability to communicate with others"

#### Chi-Square Tests

|                              | Value     | df | Asymp. Sig. (2-sided) |
|------------------------------|-----------|----|-----------------------|
| Pearson Chi-Square           | 31.658(a) | 16 | .011                  |
| Likelihood Ratio             | 35.875    | 16 | .003                  |
| Linear-by-Linear Association | 2.052     | 1  | .152                  |
| N of Valid Cases             | 141       |    |                       |

a. 16 cells (64.0%) have expected count less than 5. The minimum expected count is .34.

#### Symmetric Measures

|                                    | Value | Asymp. Std. Error(a) | Approx. T(b) | Approx. Sig. |
|------------------------------------|-------|----------------------|--------------|--------------|
| Ordinal by Ordinal Kendall's tau-b | -.125 | .074                 | -1.699       | .089         |
| N of Valid Cases                   | 141   |                      |              |              |

a. Not assuming the null hypothesis.

b. Using the asymptotic standard error assuming the null hypothesis.

Significant relationship between responses to "Most people accurately estimate how much she knows and understands" and "Apraxia (motor planning problem) limits her ability to communicate with others"

#### Chi-Square Tests

|                                 | Value     | df | Asymp. Sig.<br>(2-sided) |
|---------------------------------|-----------|----|--------------------------|
| Pearson Chi-Square              | 29.811(a) | 16 | .019                     |
| Likelihood Ratio                | 24.622    | 16 | .077                     |
| Linear-by-Linear<br>Association | 5.479     | 1  | .019                     |
| N of Valid Cases                | 141       |    |                          |

a. 18 cells (72.0%) have expected count less than 5. The minimum expected count is .13.

#### Symmetric Measures

|                                    | Value | Asymp.<br>Std.<br>Error(a) | Approx.<br>T(b) | Approx. Sig. |
|------------------------------------|-------|----------------------------|-----------------|--------------|
| Ordinal by Ordinal Kendall's tau-b | -.108 | .078                       | -1.365          | .172         |
| N of Valid Cases                   | 141   |                            |                 |              |

a. Not assuming the null hypothesis.

b. Using the asymptotic standard error assuming the null hypothesis.

## Appendix G

### Themes Expressed in Text Responses

#### The skills of girls with RS are underestimated

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"She probably knows a lot more we just haven't been able to figure it out yet"

"She clearly wants to be understood and tries to reward people with smiles and joy when they respond"

"I always knew my sister was not retarded and just had a 'block' somewhere in her communication skills. She just turned 40 and we just got a diagnosis of RS in the past 3 years. If we had known more about RS when she was younger maybe she could do more today."

"I think she knows so much more than she expresses."

"Our daughter's communication is really mostly limited by the people around here – those who make the effort to ask her opinion see that she has one, those who don't assume she doesn't know what is going on".

"She is a joy and so smart – probably smarter than we ever give her credit for".

"For some time, her teachers and therapists mistook apraxia for non-compliance. Now she has access to a number of AAC devices, modified PECS with eye gaze board, and she is really taking off."

"I think Ashley understands many more words than 50 but it is very difficult for her to communicate that she knows these words. I think she understands a great deal...especially her world around her."

"Our girl has so much to say if we only listen with our senses."

"She knows more than we think!"

"She loves people and is highly motivated to attempt communication with everyone. It really makes her very happy when others take the time to really make the effort to understand what she tries to say to them."

"The more time you spend with Tara and are able to understand her needs it makes it easier for those to work with her and give her multiple choices."

## Inconsistency of responses is a barrier to education or communication

"Due to apraxia, it is difficult for our daughter to be consistent with her responses. For example on a good day, or when she is highly motivated, she can respond within 5 seconds. On a more difficult day or time of day, it may take 30 seconds to get the same response.....the people that work with our daughter on a daily – basis – family, teacher and educational aide know and understand this, however, the consultants that work with her...the experts in OT, PT, Speech, have real difficulty because they want consistency which just isn't possible!"

"For sometime, her teachers and therapists mistook apraxia for non-compliance....now she has access to a number of AAC devices, modified PECS with eye gaze board, and she is really taking off."

"Her communication is extremely inconsistent! One day she is able to use her voice output devices to make many meaningful choices and the next day she continuously activates the non-meaningful or unpreferred picture (button)."

"Her therapist waits for her to hit the choice switch before she does what her choice is, but while she's waiting for her to hit it, Sky is staring at the picture to the point she'll almost rest her head on it before she can control her arm to hit it."

"She rarely activates high-stimulation switch toys. When presented with communication opportunities and with max prompting cues, or expectant delay, she inconsistently activates voice output communication aides....She presents with apparently low arousal levels most of her day".

"Testing her cognitive level is very difficult".

"Uses gross motor signs, repetitive hand movements sometimes get in the way of complete hand signs".

"My daughter's communication skills are inconsistent on a day to day basis. She is able to get her needs met, but I do not believe that she has an effective communication system. Even though she still uses words, sentences, pointing, eye gaze and voice activated switches, I don't believe anything is consistent enough to be effective. I strongly believe that she understands a great deal of what I say and she sees around her, but she is locked inside a body that doesn't give her a means to express consistently".

"She makes a very breathy 'yeah' sound when I ask her yes and no questions but eye gaze is her preference. Some days are much quicker and accurate than others. The apraxia is her major drawback."

"My daughter will mouth out the sounds of words like she is trying so hard to talk. This is why I know she is very smart and understands everything I say, she just can't get it out due to apraxia."

"My daughter used body language and vocal sound to express her needs. However her seizure activity seems to encompass her life. When her head is clear of seizure activity she is happy and content."

"No one in the school system would believe Sarah understood many things and Sarah became very depressed. One day after many battles we found a teacher who knew how to help Sarah communicate. After 4 years with this teacher, Sarah can multiply.....is being read chapter books and when asked questions about the chapters read she has a 96% accuracy...I strongly believe these girls can understand and do many things, given the chance."

### Strategies Used to Enhance Communication

"She used an Alpha talker with a head switch with limited success, but it was cumbersome and felt more like we were asking her to 'do tricks' than communicating. We are all much happier letting her do her thing and us 'reading' her".

"Finding the motivation and drive is the most important part of communicating with a child with Rett".

"I am starting to use switches with her. She has had them for a long time, but they only come out when I or another therapist is there. parents do not even attempt to make her use them."

"With intensive speech and ABA therapy, she began to use words, then short phrase and within 6 months was producing a constant stream of babble.....we now get at least 50 clear words/phrases (that anyone can understand) throughout the day."

"Charlotte is definitely a very visual learner and well motivated by food. She loves books and the computer. She only seems to learn when there is a strong motivator".

"Continued therapy is key. Posting pictures in black and white is helpful, ex: cracker, drink, etc. Eye gaze is key – these girls have intense eyes".

"Recent appointment with Dr. XXX raised the question of eye pointing – now we recognize she is indicating her choice/answer frequently. No wonder she was frustrated when trying to tell us something."

"She communicates mostly by body language, some vocalizations and with her eyes".

"Following introduction of Augmentative services she has added the ability to use eye gaze with picture symbols to her repertoire. She also uses switch activated items and a step-by-step sequencer".

"She shakes her head yes and no. More consistent with yes. Understands so much. Loves books and understands the story."

"She tends to take other people's hands to touch pictures or to perform tasks. She is very motivated by sounds and computers.."

"She uses an eye gaze board with up to four pictures, and looks at the picture to make a choice, and then at me to confirm. She also uses a Big Mac [switch] to participate in circle time activities. She uses a step by step sequencer to tell others what she did last night/last weekend, and when she goes home to tell her family what she did during the day. Splinting her nondominant hand increases her ability to time use of voice output more accurately".

"This young lady with RS is using a Mercury communication system. She is using direct touch access. There are some continuing issues related to some perseverative tapping around the desired item, but has made HUGE gains. She has also transitioned to all text words on her screens which contain approximately 20 to 30 items per screen using a keyguard and splints which help isolate pointer finger. She is very motivated by social interactions. Overall level of undesired behaviors has been reduced significantly!"

"We are finding that giving her the means to express herself with Mercury has lead to her showing us how much she really understands and knows...Boardmaker with Speaking Dynamically Pro was the start but now she does a lot of text only, we speak and ask her questions like anyone else and her receptive skills are age appropriate. Recognizing early on that augmentative systems was the way to open the doors of communication is important and the right kind of equipment and therapist is very important too."

"She uses a Dynavox 3100 and is amazing with it....She has limited hand use but can touch the screen to show us her wishes. She is so pleased when she succeeds in proving what she knows. She is clearly a bright little girl trapped in her rett body."